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REFERATE GENERALE /PLENARY LECTURES

ALERGOLOGIE / ALLERGOLOGY

1. ALERGIA LA ARAHIDE DE LA SIMPTOME LA PREVENTIE

Diana Deleanu
University of Medicine and Pharmacy Iuliu Hatieganu, Allergy Dept., Cluj-Napoca, Romania

Alergiile alimentare pot afecta 2-8% dintre copii. Un numar limitat de alimente, 8-14 alimente , produc 90-95% dintre simptomele de alergie alimentara, arahidele sunt dintre alimentele cel mai frecvent implicate in producerea alergiei alimentare. Prevalenta alergiei la arahide este in crestere in SUA si Europa de Vest. Alergia la arahide persista, frecvent copii nu cistiga toleranta in cursul timpului. Simptomele alergiei la arahide sunt produse de 2-3 alergeni continuti in arahide (Ara h1 si Ara h2). Simptomele sunt mediate de IgE specifice si sunt urticaria,pruritul, angioedemul, eczema, astmul, gastro-intestinale, anafilaxie. Uneori pacientii alergici pot deceda datorita anafilaxiei severe. Diagnosticul alergie la arahide se bazeaza pe dozarea IgE specifice in ser sau evidentiate prin testarea cutanata prick si pe testele de provocare. Tratamentul alergiei la arahide include evitarea contactului cu acestea. Pacientii trebuie educati sa evite alimentele ce pot contine chiar si nurme de arahide, sa aiba asupra lor trusa de urgenta ce include seringa preincarcata cu epinefrina/adrenalina. In ultimii ani diferite studii au reusit sa induca o toleranta la arahide prin imunoterapie orala cu arahide. Pentru a preveni dezvoltarea alergiei la arahide este recomandata introducerea precoce in viata la arahidelor (asa cum a demonstrat si studiul LEAP).

PEANUT ALLERGY FROM SYMPTOMS TO PREVENTION

Diana Deleanu
University of Medicine and Pharmacy Iuliu Hatieganu, Allergy Dept., Cluj-Napoca, Romania

Food allergies may affect 2-8% of the children. A few number of foods, 8-14 foods, produce 90-95% of the symptoms of food allergy, peanut being one of the most frequent allergen inducing severe symptoms. The prevalence of peanut allergy is increasing in USA and also in West Europe. Peanut allergy is a long lasting allergy (usually children do not out grow their peanut allergy). The symptoms of peanut allergy are induced by 2-3 allergen contained in peanuts (Ara h1 and Ara h2). Symptoms are mediated by specific IgE and are urticaria,itch, angioedema, eczema, asthma, gastro-intestinal manifestations, anaphylaxis. Some of the allergic patients died due to severe anaphylaxis. Diagnosis of peanut allergy is based .on specific IgE evaluation (skin prick tests, serum evaluation), food challenge. Management of the peanut allergy is avoidance and therapy of the manifestation. Patients should be educated to avoid peanut in different foods, to have their emergency kit, including auto-administrated epinephrine. In the last years different trials have succeded of inducing tolerance to peanut by oral immunotherapy. To prevent to development of peanut allergy it is recommended to introduce peanuts early in life, as LEAP study demonstrated.

2. POSIBILITĂȚI ȘI LIMITE ÎN TRATAMENTUL ECZEMEI ATOPICE LA COPIL

Stela Goția
Universitatea de Medicină și Farmacie ”Gr.T.Popa” Iași

Eczema atopică (EA) cu debut în primii doi ani de viață, corelată majoritar cu alergia (mediere IgE), se poate remite în prima decadă a vieții sub tratament comun. Totuși, 40-60% cazuri continuă să evolueze cu recurențe și agravări fiind parțial sensibile sau refractare la corticoterapie și/sau imunomodulatoare locale. EA este agravată de infecții și reprezintă un important factor de risc pentru astmul bronșic. Profilaxia EA prin măsuri pre și postnatale se menține în domeniul controverselor. Prezența EA impune o terapie holistică: nutrițională, de menajare a microbiomului intestinal, evitarea alergenilor incriminați, desensibilizări, antihistaminice și anxiolitice, susținerea barierei cutanate etc. terapia reactivă comună cuprinde glucocorticoizii topici, eventual sistemici și imunomodulatoare, în funcție de severitatea EA. Este urmată de terapia proactivă - ”terapie minimă pentru eczemă minimă”.

Heterogenitatea EA exprimată prin variate endofenotipuri și respectiv fenotipuri clinice distincte, a impus dezvoltarea abordării sistemice a inflamației cronice prin: imunosupresive și imunomodulatoare, terapii biologice, imunoterapie specifică etc. la copil tratamentele sunt limiitate de vârstă și au un risc crescut de complicații. Enigmele patogeniei limitează posibilitatea introducerii unui tratament radical al formelor severe de EA cu potențial de cronicizare.

CARDIOLOGIE / CARDIOLOGY

3. ROLUL POLIMORFISMULUI GENETIC, BIOMARKERILOR ȘI ECOGRAFIEI CARDIACE îN EVALUAREA RISCULUI DE CARDIOTOXICITATE POSTCHIMIOTERAPIE LA COPIII CU NEOPLAZII

Cainap Simona[1], Popa Gheorghe[1], Blag Cristina[1], Laura Pop[2], Cainap Calin[2], Nagy Viorica[2]
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Complicațiile cardio-vasculare reprezintă principala cauză de morbiditate și mortalitate la supravietuitorii unei neoplazii cu debut in copilarie. Childhood Cancer Survivor Study arată că riscul de deces, datorat bolilor cardio-vasculare este de 8 ori mai mare la supraviețuitorii unei neoplazii tratați cu antracicline, comparativ cu populația generală. Cardiotoxicitatea indusă de antracicline poate să apară în timpul chimioterapiei sau la scurt timp după terminarea acesteia, sau tardiv la câțiva ani după terminarea tratamentului. Identificarea precoce a pacienților cu risc de a dezvolta cardiotoxicitate reprezintă un obiectiv major, atât pentru cardiologici cât și pentru oncologi, acest lucru permițând adoptarea unor scheme de tratament personalizate, monitorizarea optimă a funcției cardiace și aplicarea unui tratament de susținere adecvat. Lucrarea isi propune analiza multidimensională a elementelor de diagnostic și prognostic: rolul mutatiilor genetice corelat cu parametrii ecocardiografici și markeri biologici. Cuvinte cheie : antracicline, cardiotoxicitate

ROLE OF GENETIC POLYMORPHISM, BIO-MARKERS AND CARDIAC ECOGRAPHY FOR EVALUATION OF CARDIOTOXICITY RISK SECONDARY TO CHEMOTHERAPY TREATMENT IN CHILDREN WITH MALIGNANCY

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Late cardiovascular toxicity represent the main cause of morbidity and mortality to the long time survivers of a childhood malignancy. Childhood Cancer Survivor Study showed an 8 fold increase of the risk of death compared with general population, for survivers of a malignancy treated with antracyclines, due to cardiovascular diseases. This cardiotoxicity could be acute – during or immediately after chemotherapy administration, or lately even after several years from the last cycle of treatment. Early prediction of cardiotoxicity represents a major issue for cardiologists and oncologists. Selecting the best candidates for chemotherapy regimens could improve the chance of a personalised therapy, optimal monitoring of cardiac function and administration of an adequate suportive treatment.
This paper will present a review of prognostic and diagnostic factors, genetic mutations correlated with echocardiography parameters and biological markers.
Keywords : antracycline, cardiotoxicity

4. AFECTAREA CARDIACA IN LIMFOAMELE MALIGNE NON HODGKIN LA COPIL

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Afectarea cardiaca in limfoamele maligne la copil, alta decat aceea indusa de tratamentul specific,survine in 5-10% din cazuri: cel mai frecvent in limfoamele maligne non Hodgkin (LNH) si poate agrava evolutia bolii.LNH cardiace: primitive: 1.3% - 2% din tumorile cardiace primitive; ·secundare: 15, 5% dintre metastazele cardiace. Manifestarile clinice de suferinta cardiovasculara sunt in general dominate de semnele clinice ale afectiunii de baza, localizarea cardiaca este relativ rar evidentiata intravitam . Ecocardiografia permite identificarea precoce a leziunilor metastatice sau primitive: · pericardica(pericardita / tamponada cardiaca-18%), revarsatul pericardic frecvent serohemoragic,citologia de tip limfomatos constituind argument major pentru diagnostic în cazurile cu dificultati de realizare a biopsiei ; · insuficienta cardiaca ;· Sindromul de vena cava superioara (SVCS), LNH-a doua cauza de neoplazie asociata cu SVCS ; · tulburari de ritm cardiac; · sincopa;· afectare valvulara primitiva sau metastatica; · boala ischemica cardiaca. Prognosticul afectarii cardiace în LNH este acela a bolii de baza. Concluzii: Examenul ecocardiografic constituie element major de diagnostic al afectării cardiace relativ frecvente în LNH la copil și trebuie efectuat în toate cazurile chiar in absența semnelor de suferinta cardiaca, in scopul prevenirr evolutiei spre afectare cardiaca severa.Ecocardiografia permite urmarirea obiectiva a evolutiei acesteia sub tratamentul specific.
Cuvinte cheie: limfom malign non Hodgkin, afectare cardiaca, ecocardiografie, copil

CARDIAC INVOLVEMENT IN MALIGNANT NON-HODGKIN LYMPHOMAS IN CHILDREN

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Cardiac involvement in malignant lymphomas in children, other than that induced by specific treatment, occurs in 5-10% of cases, most often in non Hodgk malignant lymphoma (NHL) can worsen the evolution of malignant disease. The cardiac LNH: • primary: 1.3% - 2% of the primary cardiac tumors; • Secondary: 15, 5% of cardiac metastases. Clinical manifestations of cardiovascular distress are generally dominated by clinical signs of underlying disease, cardiac involvement is rarely highlighted intravital . Echocardiography allows early identification of primary or metastatic lesions : • pericardial (pericarditis / tamponade-18% of cases), pericardial effusion frequently serohemoragic, cytology type of lymphoma being major argument for diagnosis in cases whehe the difficulty of making them biopsy; • heart failure; • Superior vena cava syndrome (SVCS), NHL-second cause of neoplasia associated with SVCS; • heart rhythm disturbances; • syncope; • valvular primary or metastatic disease; • ischemic heart disease. Heart disease prognosis in NHL is the underlying disease.

Conclusion. Because of high incidence and severity of cardiac involvement in NHL in child, echocardiography is necessary in all cases, even if cardiac involvement signs are missing, in order to prevent the evolution to severe manifestations of cardiac suffering . Echocardiography is the most important investigation to diagnose and follow-up the cardiac involvement in malignant lymphoma in child under specific treatment.

Key words: malignant non Hodgkin lymphoma, cardiac involvement, echocardiography, child

5. CARDIOMIOPATIA HIPERTROFICA IN PATOLOGIA PEDIATRICA – PROBLEME DE DIAGNOSTIC

Amalia Fagarasan

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Cardiomiopatia hipertrofica (CMH) reprezinta o boala primara a miocardului cu variate forme de expresivitate clinica, cel mai frecvent cu determinism genetic. Manifestarile clinice sunt corelate cu severitatea modificarilor hemodinamice. Din aceast motiv CMH in practica pediatrica pune serioase probleme de diagnostic, curs evolutiv si tratament. Majoritatea copiilor cu CMH sunt asimptomatici sau prezinta o simptomatologie frusta, astfel identificarea bolii se face cu ocazia unor examinari ecocardiografice de rutina. Din nefericire sunt si situatii in care prima forma de manifestare clinica a bolii este moartea subita cardiaca. In acest context este esentiala identificarea precoce a bolii inca din perioada copilariei. Cea mai mare provocare in managementul CMH ramane identificarea pacientilor cu risc pentru moarte subita cardiac. Ecocardiografia 2D reprezinta principala modalitate de screening pentru diagnostic si cuantificarea formelor clinice de CMH, permitand in egala masura evaluarea structurii si functiei cardiace. Ecocardiografia este in masura sa cuantifice gradul de severitate a CMH si sa ofere indicatori care sa ajute la alegerea modalitatilor optime de tratament: medicamentoas, chirugicala sau interventionala.Un aspect particular al acestei boli il reprezinta formele cu debut in perioada neonatala si formele de CMH secundare sindroamelor genetice.

HYPERTROPHIC CARDIOMIOPATHY IN PEDIATRIC PATHOLOGY- PROBLEMS OF DIAGNOSIS

Amalia Fagarasan

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Hypertrophic cardiomiopathy (HCM) is a primary disease of the myocardium with many clinical facets, most of which are genetically determined. The varied clinical manifestations are correlated with hemodynamic severity. For this reason, HCM is seen as a challenge in pediatric practice concerning diagnosis, clinical course and treatment. Most children with HCM may be asymptomatic or only mildly symptomatic and are often identified during routine echocardiographic examinations. Unfortunately, there are certain cases where the first clinical manifestation of the disease is sudden cardiac death. Therefore it is very important to identify the disease precociously during childhood. The most important challenge in the management of HCM is identifying those patients at increased risk for sudden death. 2D-echocardiography constitutes the primary screening tool to rule out the presence of HCM and allows a comprehensive assessment of cardiac structure and function. Echocardiography is able to quantify the hemodynamic severity of HCM, and guide the choice of therapy used: medical, surgical or interventional. A particular aspect of this disease is early onset HCM and secondary HCM linked to genetic syndromes.

ENDOCRINOLOGIE ŞI DIABET PEDIATRIC /PEDIATRIC ENDOCRINOLOGY AND DIABETES

6. CRONODISRUPTIA SI OBEZITATEA

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Creşterea alarmantă a prevalenţei obezităţii de tip "pandemic" este corelată unui spectru larg de factori, printre care cei mai des amintiţi sunt modificările majore ale compoziţiei macronutrientelor din registrul dietelor actuale si influenţele nefaste ale noului stil de viaţă marcate de sedentarism şi deprivare cronică de somn.
Ritmurile circadiene si ultradiene fac obiectul cronobiologiei. Ritmurile circadiene sunt cele care survin odata la 24 de ore fiind sincronizate luminii solare. Cel care domina toate activitatile noastre este in mod cert, ritmul somn-veghe. Cercetari recente, sustin premisa conform careia, ritmurile circadiene controleaza inclusiv modelul alimentar uman. Ca atare, *cand* mancam poate fi la fel de important cu *ce* mancam. Ingestia alimentara nocturna, deprivarea cronica de somn, "poluarea luminoasa" nocturna sunt toate responsabile de disruptia ritmului circadian sau cronodisruptie.
Cronodisruptia cu tot cortegiul sau de modificari morbide (hipercorticism, valori scazute ale STH-ului, leptinei si melatoninei, etc.) este un concept relativ nou incriminat ca responsabil de obezitate, afectare cardiovasculara, cognitiva, senescenta prematura si nu in ultimul rand, cancer.
Lucrarea de faţă sumarizează progresele înregistrate în descifrarea rolului pivot care revine cronodisruptiei in promovarea modificarilor care intervin in controlul neuroendocrin al apetitului, obezitatea fiind la ora actuala asimilata indubitabil patologiei generate de disruptia cronobiologiei normale.
Cuvinte cheie: ritmuri circadiene, cronobiologie, cronodisruptie, obezitate.

CHRONODISRUPTION AND OBESITY

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The alarming increase of so-called "pandemic" obesity is linked to a wide range of factors, among which, the most frequently reported are the major changes in the macronutrients content of the current diets and the deleterious effects of the new lifestyle marked by sedentarism and chronic sleep deprivation.
Chronobiology deals with circadian and ultradian rhythms. Circadian rhythms are those that occur once a day and are related to and synchronized with sunlight. Of course, the one that dominates all our activities is the sleep and wake cycle. Recent research supports the premise that circadian rhythms also control the eating pattern so that, *when* you eat could be just as important as *what* you eat. Night-time eating, sleep deprivation, nocturnal "light pollution", are all responsible for severe disruption of the normal circadian rhythms or chronodisruption.
Chronodisruption along with its related comorbidities (hypercorticism, low level of GH, leptin and melatonin, etc.) is a relatively new concept incriminated as being responsible for obesity, cardiovascular involvement, cognitive impairment, premature aging and last but not least, cancer.
Because obesity is undoubtedly assimilated today to the medical conditions related to the disruption of the normal chronobiology, this paper presents the pivotal role of chronodisruption in the neuroendocrine control of appetite among these patients.
Keywords: circadian rhythms, chronobiology, chronodisruption, obesity.

7. EFECTE NON-CLASICE ALE VITAMINEI D ÎN PATOLOGIA PEDIATRICĂ

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Introducere. In ultima perioadă studiile arata existenta deficitului de vitamina D la populatia pediatrică, astfel în SUA deficiența și insuficiența cumulată depășește 50% din copii și adolescenți.
Scop. Lucrarea aduce în discutie efectele non-clasice ale deficitului de vitamina D. Vitamina D influențează direct celulele T și B și implicit răspunsul lor la activare. Efectul 1,25 $(OH)_2D3$, în cazul unui antigen specific, este de a inhiba proliferarea celulelor T. Adăugarea vitaminei D la CD4 inhibă proliferarea Th1 și scade secreția de IL-2 și IFNy, iar producerea de IL-4 creste.
Studii diverse sugereaza asocierea deficit de vitamina D - dermatita atopica dar si cu alte boli alergice. Rezultate promitatoare au fost raportate în ameliorarea dermatitei atopice dupa suplimentare cu vitamina D. In aceeasi directie s-a dovedit ca un consum redus de vitamina D in timpul sarcinii creste riscul pentru astm la copiii acestor mame.
Vitamina D afectează proliferarea şi durata de viaţă a celulelor beta pancreatice fiind implicata şi in patogneza DZ tip 1. Există dovezi care sugerează rolul alterării homeostaziei vitaminei D şi Ca și în instalarea DZ tip 2. Aportul inadecvat de Ca/

vitamina D, poate altera echilibrul dintre rezervele de Ca intra- și extracelulare. Un element de legătură între insulino-rezistență și vitamina D este reprezentat de PTH, cunoscut fiind faptul că hipovitaminoza D determină o creștere compensatorie a secreției de PTH care în concentrații crescute inhibă sinteza și secreția de insulină din celulele β.
Concluzii. Determinarea anuală a 25 (OH) vitaminei D pare rezonabilă pentru monitorizarea deficitului de vitamina D.

NON-CLASSICAL EFFECTS OF THE VITAMIN D IN PEDIATRIC PATHOLOGY

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Introduction. Lately, studies continously show the existance of vitamin D defficiency in pediatric population. In the USA over 50% of children and adolescents present vitamin D defficiency and insufficiency.
Aim. This study assesses the non-classical effects of vitamin D defficiency. Vitamin D has a direct influence on T and B cells and also on their response when activated. The effect of 1,25 $(OH)_2D3$ in case of a specific antigen, is to inhibit T cell proliferation. Adding vitamin D at CD4 inhibits Th1 proliferation and decreases the IL-2 and IFNy secretion while IL-4 levels rise.
Several studies suggest the association between vitamin D defficiency and atopic dermatitis and also with other allergic disseases. Promising results regarding atopic dermatitis have been reported after administrating vitamin D supplements. It has also been demonstrated that a reduced intake of vitamin D during pregnancy rises the risk of asthma in these mother's children.
Vitamin D affects the proliferation and life duration of pancreatic beta cells, also being involved in type 1 DM pathogenessis. There are evidence that suggest the role of vitamin D and Ca homeostasys alteration also in type2 DM onset. The inadequate intake of Ca / vitamin D can alter the ballance between intra- and extracellular supplies. A linking element between insulin resistance and vitamin D is represented by the PTH, knowing the fact that vitamin D hipovitaminosys induce a compensatory growth of PTH secretion. Grown levels of PTH inhibits the synthesys and secretion of insulin from β cells.
Conclusions. 25 (OH) vitamin D annual determination seems resonable for monitoring vitamin D defficiency.

8. BOALA GRAVES – PARTICULARITATI LA COPIL

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Boala Graves (BG) reprezinta cea mai frecventa cauza de tireotoxicoza la copil si adolescent. Este produsa de stimularea imunologica a receptorului hormonului stimulator tiroidian iar incidenta la copil este redusa (1/10.000). In ciuda tratamentului de lunga durata – deseori ani – cu antitiroidiene de sinteza (ATS), remisiunea de durata se obtine doar la un numar redus de copii. Majoritatea pacientilor, necesita tratament chirurgical sau, selectiv, tratament cu iod radioactiv (RAI, I^{131}).
ATS, respectiv metimazol, carbimazol si propilthiouracil (PTU). sunt folosite cu precautie la copil, in special PTU care este asociat cu un risc inalt de insuficienta hepatica la copil, si, de aceea, este rareori folosit. Durata terapiei cu ATS, pentru obtinerea remisiunii, este mai mare decat la adult, in medie 2 pina la 5 ani. Totusi, rezultatele raportate pina acum, privind remisiunea dupa 2 ani de ATS, arata o rata redusa.
Folosirea RAI la copil este controversata, desi, numeroase studii raporteaza rezultate foarte bune dupa RAI si rate de remisiune care depasesc 95%. Totusi, trebuie avut in vedere riscul de cancer tiroidian, care dupa iradierea externa creste invers proportional cu varsta copilului si, in special la copilul sub 5 ani.
La cazurile care necesita tratament chirurgical, se recomanda tiroidectomia totala sau subtotala, pentru evitarea recaderilor. Este preferata la copilul mic (<5 ani) sau la copilul mai mare cu tiroida voluminoasa, cu raspuns slab la RAI.
Concluzii. Alegerea terapiei adecvate la copilul cu BG poate fi dificila. Pacientii cu BG trebuie monitorizati indeaproape, pentru posibilele efecte secundare ale terapiei.

GRAVES DISEASE – PARTICULAR ASPECTS IN CHILDREN

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Graves disease is the most common cause of thyrotoxicosis in children and adolescents. It is caused by an immunologic stimulation of the thyroid stimulating hormone receptor and the incidence in children is low (1/10.000). Despite long term treatment – usually for many years - with antithyroid drugs (ATDs), long lasting remission occurs only in a minority of treated children. Most patients will need surgical treatment or, in some selected cases, treatment with radioactive iodine (RAI, I^{131}).
ATDs including methimazole, carbimazole and propylthiouracil (PTU) are used with precaution in children, especially PTU, which is associated with a high risk of severe liver injury at this age group, thus, is rarely used. ATD therapy is usually longer

than in adults, 2 to 5 years, to obtain remission. However, to date reported remission rates, after 2 years of ATDs treatment, are low.
The use of RAI therapy is controversial, although studies focused on its use in children reported excellent outcomes and remission rates exceeding 95%. Still, it should be kept in mind that the risk of thyroid cancer, after external irradiation, increases the younger the child is, especially in children younger than 5 years.
When surgical treatment is needed, total or near total thyroidectomy is recommended, to avoid relapse. It is preferred in young children (<5 years) and in older children with large thyroid with poor response to RAI.
Conclusions. Choosing the proper therapy for a child with GD may be difficult. Patients should be closely and carefully monitored for side effects.

GASTROENTEROLOGIE / GASTROENTEROLOGY

9. ABORDAREA DIAGNOSTICA SI TERAPEUTICA A BOLII INFLAMATORII INTESTINALE CU DEBUT FOARTE PRECOCE

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Relatia dintre varsta la care se stabileste diagnosticul si evolutia bolii inflamatorii intestinale (BII) pediatrice nu este bine cunoscuta. Totusi este evident ca BII are particularitati la varsta pediatrica si pentru prima data, clasificarea Montreal a delimitat, ca grup distinct, pacientii cu debut sub varsta de 17 ani (BII cu debut la varsta pediatrica).
In prezent se propune delimitarea unor subgrupe de BII pediatrica in functie de varsta: BII cu debut pediatric (<17 ani), BII cu debut precoce (<10 ani), BII cu debut foarte precoce (<6 ani), BII cu debut infantil (0-2 ani), BII neonatala (<28 zile)
S-a raportat creşterea numărului pacienţilor pediatrici cu BII şi debutul bolii la vârste din ce în ce mai mici.
Au fost identificaţi 163 loci genetici asociaţi cu susceptibilitatea pentru BII.
Tendinţa BII de a debuta precoce este determinate de predispoziţia genetică si de modificările factorilor de mediu.
Până în prezent sunt cunoscute 50 defecte monogenice asociate cu patologie BII-like
Istoricul bolii, examenul clinic, endoscopia digestiva, investigatiile imagistice sunt necesare pentru a stabili diagnosticul de BII. Infectiile enterale, frecvente la varsta pediatrica, trebuie sa fie excluse prin examene de laborator. Alergia la proteinele din laptele de vaca si boala celiaca vor fi de asemenea excluse.
S-a demonstrat ca BII cu debut foarte precoce are o incidenta in crestere si un numar din ce in ce mai mare de cazuri cu colita ulcerative cu activitate severa.
BII cu debut foarte precoce are un prognostic sever si adesea necesita o abordare terapeutica agresiva.

DIAGNOSIS AND THERAPEUTIC MANAGEMENT OF VERY EARLY ONSET INFLAMMATORY BOWEL DISEASE

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The relation between the age at diagnosis and the evolution of pediatric inflammatory bowel disease is not well known. However it is obvious that IBD has particular characteristics in pediatric patients.
For the first time, Montreal classification delimited patients whose disease started under age 17, as a distinct group of patients – pediatric IBD. Recently there were proposed IBD subgroups according to age: very early onset IBD - (<6 years), infantile IBD (<2 years) and neonatal IBD (<28 days)
An increasing incidence of IBD in pediatric patients was noticed; affected children are increasingly younger.
Over 163 associated genetic loci have been identified. This could be explained by a particular genetic predisposition to develop IBD, and also by changes in environmental factors that initiate the disease. There are at least 50 monogenic defects responsible for an IBD-like pathology
Patient history, physical examination, endoscopic investigations, imaging, are required to establish the diagnosis of IBD. Gastrointestinal infections are frequent in pediatric patients and are excluded by stool culture and virology tests. Cow's milk protein allergy and celiac disease should be excluded.
Several studies reported an increasing incidence of very early onset-IBD, frequent pancolonic ulcerative colitis, children presenting with severe ulcerative colitis disease activity
Very early IBD has a severe prognosis and often needs an aggressive therapeutic approach.

10. ALERGIA IG E DEPENDENTA LA GRAU VERSUS BOALA CELIACA

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Alergenii la grau sunt numerosi – unii solubili in apa (albumine, globuline), altii insolubili (gliadine, aglutenine). Gliadinele (30% din proteine grau) sunt alfa, beta, gama si omega gliadine (omega 1-5) dupa migrarea electroforetica, iar gluteninele reprezinta 35% dintre proteine.

Alergenicitatea acestor diverse proteine este variabila dupa individ.

Graul este unul dintre cele 6 alimente implicate in 90% dintre alergiile alimentare IgE dependente la copil.

Simptomele si semnele alergiei la grau sunt numeroase si apar la orice varsta cu tablouri particulare cunoscute ca: a) alergii profesionale (astmul brutarului); b) alergii IgE dependente – cu simptome ca: urticarie, angioedem, rinita, astm, anafilaxia; c) anafilaxii induse de exercitiul fizic si ingestia de alimente; d) alergia la izolate de grau; e) dermatita atopica si alergia la grau; f) manifestari digestive cronice la sugar si copilul mic. Aceasta varietate este in raport cu multitudinea alergenilor de grau.

Evolutia este cel mai adesea rezolutiva, dar exista si forme fixate care necesita regim de excludere sau inductia tolerantei.

Boala celiaca este o enteropatie inflamatorie cronica autoimuna declansata de ingestia de gluten la indivizi predispusi genetic.

Boala celiaca nu este o simpla enteropatie, ci un spectru clinic mai vast – este o boala sistemica. Autoimunitatea este bine demonstrata dupa descoperirea autoantigenului transglutaminaza 2 (TG2). TG2 este o enzima care provoaca deamidarea gliadinei ingerate si demasca astfel epitopii acestor proteine; ea se gaseste in ficat, rinichi, plaman, intestin, capsula articulara – fapt care ar explica manifestarile sistemice.

Ingestia de gluten (gliadina) este factorul declansant al bolii. Expunerea la gluten inainte de varsta de 3 luni mareste riscul de 2-3 ori, iar peste 7 luni – mareste riscul de 4 ori.

Se stipuleaza ca exista o fereastra de oportunitate pentru introducerea glutenului in alimentatie intre 4-7 luni, iar continuarea alimentatiei naturale in cursul diversificarii reduce riscul de boala celiaca. Fainoasele interzise sunt secara (secalina), graul (gliadina), orzul (hordeina).

Orezul si porumbul nu sunt toxice pentru bolnavul celiac si sunt folosite ca substitut in regimul fara gluten.

Boala celiaca are un teren genetic predispozant: 8% dintre bolnavi exprima HLA DQ2 si / sau DQ8.

Modalitatile de diagnostic s-au schimbat mult, reprezentand un adevarat seism. Practica dozarii autoanticorpilor in boala celiaca (antigliadine, antiendomisium si anti transglutaminaza IgG sau IgA) a permis limitarea biopsiilor intestinale, fiind stabilit in prezent un algoritm precis de diagnostic la copii simptomatici si la cei asimptomatici, elaborat de ESPGHAN.

IGE DEPENDENT WHEAT ALLERGY VS. CELIAC DISEASE

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Wheat allergens are numerous - some are water soluble (albumin, globulins), others insoluble (gliadin, aglutenine). Gliadins (30% of wheat proteins) are alpha, beta, gamma and omega gliadin (omega 1-5) by electrophoretic migration and glutenins are 35% of the proteins. Allergenicity of these various proteins varies by individual.

Wheat is one of the six foods involved in 90% of IgE-dependent food allergy in children. Wheat allergy symptoms and signs are numerous and occur at any age. Some are known as: a) occupational allergy (baker's asthma); b) IgE-dependent allergies - with symptoms such as urticaria, angioedema, rhinitis, asthma, anaphylaxis; c) exercise-induced anaphylaxis and food intake; d) isolated allergy to wheat; e) atopic dermatitis and allergy to wheat; f) chronic digestive manifestations in infants and young children. This variety is in relation to the multitude of wheat allergens. Evolution is often towards resolution, but there are forms that require fixed exclusion regime or tolerance induction.

Celiac disease is an autoimmune enteropathy triggered by chronic inflammatory ingestion of gluten in genetically predisposed individuals. Celiac disease has a broader clinical spectrum –it is a systemic disease. Autoimmunity is well demonstrated by the discovery autoantigen transglutaminase 2 (TG2). TG2 is an enzyme that causes deamidation of ingested gliadin epitopes of these proteins; it is found in liver, kidney, lung, intestine, joint capsule - which would explain systemic manifestations.

Ingestion of gluten (gliadin) is causing the disease. Exposure to gluten before 3 months of age increases the risk by 2-3 times, and introduced at more than seven months - increases the risk by 4 times.

It stipulates that there is a window of opportunity for the introduction of gluten in the diet between 4-7 months while continuing complementary feeding; natural nutrition reduces the risk of celiac disease. Prohibited foods are rye (secalin), wheat (gliadin), barley (hordein). Rice and corn are not toxic to celiac pacient and are used as substitute for gluten-free diet.

Celiac disease has a predisposing genetic terrain: 8% of patients express HLA DQ2 and / or DQ8.

Diagnostic modalities have changed lately, representing a real earthquake. Practice of dosing autoantibodies in celiac disease (antigliadine, antiendomisium IgG or IgA and anti transglutaminase) allowed limitation of intestinal biopsies; currently there is a well established diagnostic algorithm in children (both symptomatic and asymptomatic), developed by ESPGHAN.

11. .TRATAMENTUL BOLII CELIACE, AZI, MÂINE!?

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Consiliere dietetica si dieta STRICT fara gluten
La inceput, este indicata evitarea consumului de produse lactate
Intoleranta la lactoza secundara
Inlocuirea micronutrientilor in caz de deficienta
70% din pacienti dovedesc imbunatatire clinica in doua saptamani
Dar: simptomele persista la pana la 20% si pana la 2-3% din pacientii varstnici nou diagnosticati dezvolta boala celiaca refractara
Evitarea stricta a tuturor surselor (inclusive cele ascunse) de gluten in dieta
Regim fara gluten =< 20mg/kg
Simptomele caracteristice bolii celiace:

- Glutenul din faina de grau, orz si secara declanseaza imunitatea (celule T)
- Predispozitie genetica (HLA-DQ2 sau DQ8)
- Autoanticorpii transglutaminaza de tip IgA

Nevoia se terapie farmaceutica:

- Pentru a trata pacientii care sunt excesiv de sensibili la urmele de gluten (RCD)
- Pentru a permite pacientilor o dieta fara gluten mai putin stricta (evitarea surselor de gluten evidente)
- Pentru a permite transgresiuni dietetice, de ex. in cazul calatoriilor
- Pentru o posibila recapatare a tolerantei fata de gluten („insanatosire’’)

Creste degradarea glutenului:

- AN-PEP: endoproteaza a Asperggilus niger
- Descompune peptidele si proteinele
- Functioneaza la pH redus, rezistent la pepsina
- Degradeaza glutenul sub conditii gastrointestinale simulate
- Se poate produce in cantitate mare.

Stepniak et al, 2006; Mitea et al, 2008
Tulburari ale IL-15/IL-15R alfa in boala celiaca: implicatii pentru imunoterapie

- IL-15 este presenta in cantitati extreme de mari in lamina propria si in epiteliul intestinal la pacientii cu boala celiaca refractara
- IL-15 induce proliferarea si marirea inmultirii anormale intraepiteliala
- Aceste observatii sugereaza folosirea Hu-Mik-beta-1, care blocheaza actiunea IL-15 in tratamentul pacientilor cu boala celiaca refractara
- Sunt initiate testari de Hu-Mik beta-1 (anti-IL-2/IL-15 receptor, CD 122) intr-o Faza II a testarii open-label a 10 pacienti cu boala celiaca refractara

Elafina, moduleaza inflamatia la nivelul colonului

- “Am studiat rolul elafinei in boala celiaca, folosind tesut intestinal uman de dimensiuni mici in vitro. De asemenea, am studiat potentialele efecte benefice ale elafinei intr-un model al sensibilitatii glutenului pe soareci

Aparitia elafinei in epiteliile intestinale subtiri a fost mai redusa la pacientii cu boala celiaca activa decat la pacientii aflati sub control”

TRATMENT OF CELIAC DISEASE, TODAY, TOMORROW!?

Alexandru Pirvan
UMF Cluj-Napoca

Dietary consult and STRICT gluten free diet
At the onset it may be useful to avoid excess milk products (secondary lactose intolerance)
Replacement of micronutrients in case of deficiency
70% of patients show clinical improvement within 2 weeks
But: complaints persist in up to 20 % and up to 2-3% of newly diagnosed elderly adults may develop refractory celiac diseaseGluten-free diet (GFD) Strict avoidance of all sources of (hidden) gluten in the diet : Gluten-free = < 20 mg/kg
Hallmarks of celiac disease

- Dietary gluten from wheat, barley, or rye as trigger of adaptive (Tcell) immunity
- Genetic predisposition (HLA-DQ2 or-DQ8)
- IgA autoantibodies to tissue transglutaminase
- A wheat component that drives innate immunity

Need for pharmacological therapy

- To treat patients who are excessively sensitive to traces of gluten (RCD)
- To permit patients a less strict gluten free diet (avoiding only obvious sources of gluten)
- To allow occasional dietery transgressions….during travel
- To possibly restitute tolerance towards gluten ("healing")

Enhance gluten degradation ?
AN-PEP : a prolyl endoprotease from Asperggillus niger
Cuts after proline; cuts peptides and proteins
Works at low pH, resistent to pepsin
Degrades gluten under simulated gastrintestinal conditions
Mass production in fermentors
Stepniak et al, 2006; Mitea et al, 2008
Disorders of IL-15/IL-15R alfa in Celiac disease : implications for immunotherapy

- IL-15 is massively over-expressed in the lamina propria and the intestinal epithelium in patients with refractory celiac disease.
- IL-15 induces the proliferation and augments the action of clonally abnormal intraepithelial.
- These observations support the use of Hu-Mik-beta-1 that blocks IL-15 action in the tratament of patients with refractory celiac disease.

12. . INFECTIA CU HELICOBACTER PYLORI LA COPIL

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Infecția cu *Helicobacter pylori* (*H. pylori*) este o verigă patogenică foarte importantă la copiii cu afecțiuni gastroduodenale. Rolul bacteriei *H. pylori* în patogeneza bolilor digestive este foarte bine stabilit şi cunoscut în prezent, însă ultimele cercetări argumentează implicarea *H. pylori* și în producerea unor afecțiuni extradigestive. Prevalenţa infecţiei diferă de la o zonă geografică la alta, fiind în declin în ţările dezvoltate şi menţinându-se încă ridicată la populaţia din ţările în curs de dezvoltare. Se ştie că există bacterii ce afectează specific un anumit organ, dar în același timp pot determina modificări sistemice la nivelul întregului organism. Infecția cronică cu *H. pylori* se asociază cu anemie, scădere ponderală și staturală. De asemenea, *H. pylori* este implicată în statusul pacienților diabetici și în alergiile alimentare. Creșterea calității și acurateței depistării infecției cu *H. pylori* determină o rată scăzută a complicațiilor acestor afecțiuni prin aplicarea unor strategii de tratament constant îmbunătățit.

Cuvinte cheie: *Helicobacter pylori,* afecțiuni digestive, afecțiuni extradigestive, copil

INFECTION WITH HELICOBACTER PYLORI IN CHILDREN

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The infection with *Helicobacter pylori (H. pylori)* is a very important pathogenetic link in children with gastroduodenal diseases. The role of *H. pylori* bacteria in the pathogenesis of digestive diseases is well established and currently known, but recent research argues the involvement of *H. pylori* in the occurrence of extradigestive diseases. The prevalence of infection differs from one geographical area to another, being in decline in developed countries and keeping still high among the population of developing countries. It is known that there are specific bacteria that affect a particular organ, but also can cause systemic changes throughout the body. Chronic infection with *H. pylori* is associated with anemia, loss of weight and low stature. Also, *H. pylori* is involved in diabetic patients status and food allergies. Increasing the quality and accuracy of detection of infection with *H. pylori* causes a low rate of complications of these diseases by using constantly improved treatment strategies.

Keywords: *Helicobacter pylori,* digestive disease, extradigestive diseases, child

13. DE CE ESTE DIFICILA TERAPIA BOLII CROHN PEDIATRICE?

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Incidenta bolii Crohn (BC) este in continua crestere, mai ales in zone unde inainte aceasta era neobisnuita. In mod particular, terapia copiilor cu BC poate fi foarte problematica, intrucat aspectele de debut si evolutia sunt mai severe decat la adult. Scopul principal al terapiei actuale este obtinerea remisiei profunde, cu vindecarea mucoasei (ideal si cu normalizare transmurala). In ciuda progreselor enorme in dezvoltarea de noi medicatii, BC nu este deocamdata curabila si nu exista nicio terapie cu succes de 100%. Pacientii si familiile acestora trebuie informati de la inceput despre impredictibilitatea evolutiei BC. Diagnosticul tardiv poate afecta succesul terapiei, pacientii avand deja forme complicate de BC. Mijloacele medicale

disponibile actual (nutritia enterala exclusiva – NEE, corticoizii si agentii anti-factor de necroza tumorala – TNF) pot fi eficace in inductia remisiei clinice la majoritatea pacientilor, dar vindecarea mucoasei este obtinuta mai ales prin EEN si agenti anti-TNF. Pentru prevenirea puseelor, imunomodulatoarele si agentii anti-TNF pot fi eficace, dar remisia profunda pe termen lung este mentinuta la mai putin de 50% dintre pacienti. Multi copii inca necesita interventii chirurgicale si nu exista o terapie eficace in prevenirea recidivei post-interventionale. Majoritatea mijloacelor terapeutice au reactii adverse, de aceea beneficiile si riscurile trebuie intotdeauna discutate cu pacientii si familiile lor. Alte probleme terapeutice care trebuie avute in vedere sunt: corectarea deficitelor nutritionale, cu restaurarea cresterii (inclusiv osoase) si dezvoltarii, ameliorarea calitatii vietii si maximizarea aderentei la terapie. Prezentam dificultatile de tratament ale bolii Crohn, cu exemplificari din experienta Clinicii Pediatrie II Cluj-Napoca (1998-2015).

WHY IS CROHN'S DISEASE DIFFICULT TO TREAT IN PEDIATRICS?

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Incidence of Crohn's disease (CD) continues to rise, especially in regions where previously it was not common. Particularly in children, therapy of this condition could be very problematic, as disease presentation and course are more severe than in adults. Nowadays, deep remission (with mucosal - and preferably - transmural healing) represents the major therapeutic goal. Despite enormous progress in development of new drugs, there is yet no cure for CD and no therapy with 100% success. Patients and families should be informed from the beginning about the unpredictability of CD course. Belated diagnosis could impair a succesful therapy, patients presenting with already severe formes of CD. Available medical tools [i.e., exclusive enteral nutrition - EEN, corticoids, anti-tumour-necrosis-factor (TNF) agents] may work well in induction of clinical remission in most patients, however mucosal healing could be obtained especially with EEN and anti-TNF. For prevention of relapses, immunomodulators and anti-TNF could be useful, but long-term deep remission is achieved in less than 50% of patients. Many patients still require surgery and there is no effective therapy to prevent post-surgical recurrence. Most therapies have adverse reactions and benefits and risks should be always discussed with patients and families. Other therapeutic issues that have to be taken into consideration are correction of nutritional deficits, with restoring growth (including bone growth and density) and development, improvement of quality of life and maximization of adherence to therapy. We discuss difficulties in treating CD children, presenting cases from the 2nd Department of Pediatrics, Cluj-Napoca (1998-2015).

14. COLONOPATIA FIBROZANTĂ DIN MUCOVISCIDOZĂ (FIBROZA CHISTICĂ) – CU ADEVĂRAT O PROBLEMĂ?

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Colonopatia fibrozanta (CPF) din mucoviscidoza - MV (fibroza chistică) reprezinta o entitate tradusă prin hipertrofia muscularei si strictură implicită a colonului ascendent. Reprezintă o particularitate de evoluție în MV și a fost descrisă pentru prima dată în 1994. De-a lungul timpului a suscitat interes deosebit, cel puțin prin faptul ca în etiologie au fost incriminate dozele excesiv de mari de enzime pancreatice (în special lipaze) și implicit riscul chirurgical. De aici problema restricției dozelor de enzime pancreatice, de multe ori în contradictie cu situațiile de insuficiență pancreatică, care necesitau doze substitutive mari. În acest sens, clasic, se recomandă ca doza de enzime pancreatice să nu depașească 10 000 UI/kgcorp/zi. Sau 2500UI/kgcorp/doza.Simptomele inițiale pot fi similare cu cele din sindromul de obstucție intestinală distala (SOID). Diagnosticul se pune prin clismă de contrast. In timp, s-a observat că pericolul legat de excesul de enzime pancreatice în tratamentul bolnavilor cu MV, nu este pe masura atenționărilor inițiale. Lucrarea realizează o trecere în revista a datelor actuale privind CPF și a recomandărilor care derivă, legat de administrarea substitutivelor enzimatice.

FIBROSING COLONOPATHY IN CYSTIC FIBROSIS- TRULY A PROBLEM?

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Fibrosing colonopathy (FCP) is an entity that arises in patients with cystic fibrosis (CF). It consists of hypertrophy of the colon muscularis and ascending colon stricture. It is a particular form of evolution in CF and was first described in 1994. Over time it has aroused particular interest, at least by the fact that the incriminated etiology was excessive doses of pancreatic enzymes (especially lipases) and because of the surgical risk it carries. Hence the problem of pancreatic enzymes restriction doses, often in contradiction with the pancreatic insufficiency seen in clinical practice, requiring large substitution doses. Thus, it is recommended that pancreatic enzyme substitution not to exceed 10000 IU / kg / day or 2500 IU/ kg / dose. Early symptoms may be similar to the distal intestinal obstructive syndrome (DIOS). Diagnosis is established via contrast enema. It was observed that the risk of excess pancreatic enzymes during the treatment of CF patients was not as high as the initial warnings assumed. This paper provides a review of the current data regarding FCP and of the enzymatic substitution recommendations.

15. .IMUNITATEA MUCOASEI TUBULUI DIGESTIV- ACTUALITĂȚI

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Intestinul este cel mai mare organ limfoid al corpului, dar și cel mai mare colecționar de microbi al organismului. Aici domnește o stare de inflamație controlată. În imunitatea digestivă sunt implicate sistemele imun înnăscut și cel adaptativ, microbiota, bariera de mucus și celulele epiteliale. Bariera mecanică a fost descifrată, cu identificarea complexelor care mențin spațiile interepiteliale: desmozomi, joncțiuni strânse, zonula adherens și moleculele proteice de adeziune. Dintre actorii imunității înnăscute tot mai bine studiați sunt TLRs (toll like receptors), unele proteine intracelulare, care recunosc patogenii intracelulari: NOD1 (nucleotide oligomerisation domain 1) şi NOD 2, celulele dendritice și macrofagele intestinale. S-a demonstrat că în boala Crohn există o secreție inadecvată de citokine de către macrofage și imposibilitatea de a realiza un clearence microbian eficient. Dintre participanții la sistemul imun adaptativ al intestinului azi se vorbește de răspunsuri imun de tip 1,2 și 3, legate de participarea limfocitului T helper Th1, Th2, respectiv Th17. La pacienții cu boală inflamatorie intestinală există interacțiuni anormale între micobiotă și sistemul imun la mucoasei. Menținerea ecosistemului intestinal în stare optimă prin modulare cu dietă și probiotice ar avea efecte benefice privind obezitatea, alergia, bolile autoimune, intestinul iritabil. Un subiect pasionant e axa-intestin creier. Microbiota intestinală are rol în procese neurohormonale, influențează funcțiile cognitive, sociale și reacțiile la stress. Terapiile actuale se concentrează pe imunitatea adaptativă și pe citokinele inflamatorii. Terapiile viitorului vor ținti spre actorii imunității înnascute, spre mecanismele de întărire a barierei mucoasei și a joncțiunilor intercelulare, spre restabilirea barierei de mucus și modularea microbiotei. Cuvinte cheie: toleranţă orală, imunitate, intestin, alergie alimentară, celiachie, boala Crohn

THE GUT IMMUNITY – AN UPDATE THE GUT IMMUNITY – AN UPDATE

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The intestine is the largest lymphoid organ, but also the largest collector of microbes. Here reigns a state of controlled inflammation. The gut defense system involves the innate and the adaptative immunity, the microbiota, the mucus barrier and the epithelium. The mechanical barrier has been deciphered, including the structures that keep tight the intercellular spaces: desmosomes, tight junctions, zonula adherens and adhesion protein molecules. The main actors of the innate immunity are: the TLRs (Toll like receptors), some intracellular proteins that recognize intracellular pathogens: NOD1 (nucleotide oligomerisation domain) and NOD 2 , the intestinal macrophages and dendritic cells. In the Crohn's disease there is an inadequate secretion of cytokines by macrophages with an ineffective microbial clearance. Regarding the adaptive immune system of the gut three types of immune responses are recognized: 1,2 and 3, involving the T helper lymphocyte Th1, Th2, Th17 respectively. In the inflammatory bowel disease there are abnormal interactions between the immune system and the microbiota. The maintenance of the intestinal ecosystem in optimal conditions by modulation of the diet and with probiotic supplementation may be beneficial in obesity, allergies, autoimmune diseases, irritable bowel. A fascinating subject is the brain-gut axis. The intestinal microbiota has a role in the neurohormonal processes influencing the cognitive, social functions, and stress management. The current therapies focus on adaptive immunity and inflammatory cytokines. The therapies of the future will target the innate immunity actors, to strengthen the mechanisms of defense of the mucosal barrier and to modulate the microbiota.

Key words: oral tolerance, immunity, gut, food allergy, celiac disease, Crohn

GENETICĂ / GENETICS

16. EVALUAREA COPILULUI CU INTARZIERE IN DEZVOLTAREA PSIHOMOTORIE SAU RETARD MENTAL

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Intarzierea globala in dezvoltare/ retardul mental este una dintre cele mai frecvente dintre cauzele pentru care este solicitata evaluarea neuropsihiatrica pediatrica. Prevalenta acesteia in populatia pediatrica nu este inca complet cunoscuta,

estimandu-se a fi situata in intervalul 1-5%, in studiile prezentand frecvente mai reduse fiind excluse tulburarile de spectru autist.

Etilogia intarzierii globale in dezvoltare/ retardului mental este foarte heterogena, faptul ca multe dintre cauze sunt actual tratabile accentuand necesitatea identificarii si diagnosticului precoce. Mai mult, pentru patologiile cu transmitere genetica se poate estima prognosticul, oferi consiliere familiala si diagnostic prenatal atunci cand este posibil.

Supravegherea dezvoltarii este recunoscuta a fi parte integranta a ingrijirii pediatrice. Desi protocolul de evaluare al copilului cu intarziere in dezvoltarea psihomotorie nu este inca definit complet, asociatiile profesionale din domeniu recomanda monitorizarea dezvoltarii pshomotorii ca practica pediatrica de rutina. Asfel, folosirea instrumentelor de screening formal impreuna cu metodele de raportare parentala sunt considerate modelul de identificarea primara a copiilor cu intarziere in dezvoltare. In plus se insista pe generarea unor programme individualizate de evaluare periodica a dezvoltarii petru copii cu risc biologis si/sau social pentru retard in achizitii.

Concluzia review-urilor recente avand scopul de a genera ghiduri de diagnostic etiologic este ca la finalul evaluarii majoritatea cazurilor pot obtine un diagnostic etiologic. Ghidurile indica testele necesare pentru diagnostic de rutina pentru ca, in functie de istoric si rezultatele examenului clinic, sa se recomande extinderea evaluarii cu analize specific.

THE EVALUATION OF THE CHILD WITH MENTAL RETARDATION OR DEVELOPMENTAL DELAY. A TEAM IS NEEDED!

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Global developmental disabilities (GDD) are one of the most frequent referral reasons in pediatric neuro-psychiatry. The prevalence of GDD in the pediatric population is not yet precisely known; however, estimates of affected children range between 1% and 5%, the lower estimated frequencies exclude the autistic disorders. Etiologies of GDD are highly heterogeneous; many of the possible causes for the clinical picture of GDD are currently treatable, this emphasizing the need for early recognition and diagnosis. In addition, for the genetically transmitted etiologies the prognosis can be determined, creating the conditions for family counseling and prenatal diagnosis when possible.

Developmental surveillance is recognized as an integral component of paediatric care. Although the optimal approach for workup of a child with GDD is not completely defined, professional organizations dedicated to the medical care of children recommend routine monitoring of a child's developmental progress. The availability of formal screening together with reliance on parental reporting measures constitutes the primary means by which children with global developmental delay are identified. It is also considered important the design of special follow-up programs that incorporate routine periodic assessments evaluating developmental performance of children with either biologic or social risk factors for later developmental delay.

The conclusion of the latest literature reviews is that a specific etiology can be determined in the majority of children with global developmental delay. There are indicated routine screening tests and, depending on history and examination findings, the evaluation will be extended with additional specific testing.

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17. TULBURARILE DE STOCARE ALE ACIDULUI SIALIC

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Acidul sialic face parte dintre rezidurile zaharice cu legarea cea mai distala atasata multor glicoproteine; este in principal prezent in sistemul nervos confirmand astfel importanta sa pentru dezvoltarea creierului.

In patologia pediatrica au fost descrise doua tulburari de stocare a acidului sialic avand model de transmitere autosomal recesiv: boala de stocare infantila a acidului sialic si boala Salla. Ambele tulburari sunt cauzate de mutatii in transportorul lizozomal de acid sialic SLC17A5 ducand la acumularea acidului sialic liber in lizozomi. Pacientii cu boala de stocare infantila a acidului sialic cel mai frecvent decedeaza in copilarie in timp ce boala Salla are un fenotip mai atenuat, fiind mai severa la adult.

In sialurie, o alta tulburare din acest spectru avand model de transmitere autosomal dominant, pacientii au mutatii ale genei pentru enzima UDP-N-acetilglucozamin 2-epimeraza/N-Acetilmanozamin kinaza care cauzeaza acumularea de acid sialic liber in celule. Acumularea libera intracitoplasmatica cauzeaza un fenotip sever consistand in retard mental, macrocefalie, hepatomegalie si dismorfism facial cu trasaturi facile accentuate.

Studiile noastre recente au pus bazele unei potentiale solutii terapeutice in sialurie. Astfel, utilizand tehnologia de ARN interferenta cu specificitate alelica pentru a silentia expresia alelei mutante in fibroblastii obtinuti de la pacienti cu sialurie, am reusit reducerea nivelurilor de acid sialic liber. Prezentarea mea se doreste un ghid pentru noile date diagnostic si de cercetare in domeniul acidului sialic si tulburarilor asociate.

SIALIC ACID STORAGE DISORDERS AND RELATED DISEASES

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Sialic acid represents the outermost residue of sugar chains that are attached to many glycoproteins; is mainly present in the nervous system, in this way confirming its importance for the brain development.

In pediatric pathology there have been described two sialic storage disorders with an autosomal recessive pattern of inheritance: Infantile Sialic acid Storage Disease and Salla disease. Both disorders are caused by mutations in the lysosomal sialic acid transporter SLC17A5 leading to accumulation of free sialic acid in lysosomes. Patients with Infantile Sialic acid Storage Disease usually die in infancy, whereas Salla disease has a milder phenotype being usually more severe at adult age.

In yet another rare disorder called Sialuria, patients have autosomal dominantly inherited mutations in the enzyme UDP-N-acetylglucosamine 2-epimerase/N-Acetylmannosamine kinase leading to an accumulation of free sialic acid in cells. The accumulation of cytoplasmic free sialic acid results in a severe phenotype consisting of mental retardation, macrocephaly, hepatomegaly, and coarse facial features.

Not long ago we have set the stage for a potential therapeutical solution of Sialuria. To this end we used allele-specific RNA interference technology to silence the expression of the mutant allele in fibroblasts obtained from Sialuria patients, leading to decreased levels of free sialic acid. During my talk I will guide you through a journey into the latest diagnostic and academic findings about sialic acid storage disorders and related diseases

18. DIABETUL ZAHARAT DE TIP 2: GENETIC SAU EPIGENETIC?

Prof. Sorin Buzinschi
Universitatea Transilvania- Facultatea de Medicina Brasov

Raportul dintre conditionarea genetica si nongenetica a diabetului zaharat de tip 2 (DZ 2) a suscitat opinii divergente in cadrul epidemiei actuale de diabet. Exceptand diabetul monogenic MODY, componenta poligenica a bolii acopera numai 10% din factorii mosteniti ai bolii. Aceasta a pus sub semnul intrebarii cuantumul componentei genetice caci fondul genetic populational se schimba foarte lent. Numeroase studii au evidentiat legatura dintre tulburari din perioada fetala, a primilor ani de viata si modificari metabolice ulterioare ca DZ 2 si sindromul metabolic. Rezistenta la insulina, marker esential al dezvoltarii diabetului, a fost pusa in evidenta la nou nascutii prematuri si la alti copii cu restrictie de dezvoltare intrauterina. Deasemeni ea afecteaza nascutii macrosomi ai mamelor obeze sau diabetice, copii care detin un procent cu 20% mai ridicat de grasime corporala fata de cei normoponderali. Datele arata ca traectoria de risc pentru DZ 2 incepe foarte devreme, din viata intrauterina. In centrul tulburarilor metabolice din DZ 2 si obezitate se afla disfunctia mitocondriala. Atat la subiectii diabetici cat si la urmasii rezistenti la insulina ai acestora s-a constatat scaderea captarii celulare de glucoza, reducerea fosforilarii mitocondriale, diminuarea sintezei de ATP si cresterea continutului de grasimi a miocitelor. Reprogramarea mitocondriala are la baza modificari de metilare ale ADN si tulburari functionale ale histonelor. Modificarile epigenetice implica metilarea regiunii promoter a genei PPARGC1A care codeaza pentru PGC-1α, pentru genele fosforilarii oxidative (OXPHOS) si pentru gena insulinei, in insulele pancreatice.

TYPE 2 DIABETES: GENETIC OR EPIGENETIC?

Prof. Sorin Buzinschi
Transilvania University- Faculty of Medicine

The relationship between genetic and epigenetic conditioning of type 2 diabetes aroused disagreements regarding the current diabetes epidemia. Excepting monogenic MODY diabetes, polygenic component of the disease covers only 10% of inherited factors. This queried the amount of genetic component in population because the genetic matrix changes very slowly. Numerous studies have shown the link between impaired fetal period, from the first years of life, and subsequent metabolic changes as type 2 diabetes and metabolic syndrome. Insulin resistance, as an essential marker of diabetes development was highlighted in premature infants and other children with intrauterine growth restriction. Also it affects infants of obese or diabetic mothers, which have a 20% higher percentage of body fat than those of normal weight. The data show that the risk for type 2 diabetes path begins very early, in fetal life.

At the core of metabolic disorders inherent to obesity and type 2 diabetes lies mitochondrial dysfunctions. For both diabetic subjects and also their descendants that have insulin resistance, decreased glucose intake, reduced mitochondrial phosphorylation, decreased ATP synthesis and increased fat content of myocytes cells was found.

Mitochondrial reprogramming is based on DNA methylation and functional disorders of histones. Epigenetic changes involve methylation of PPARGC1A gene that is coding for PGC-1α, for oxidative phosphorylation genes (OXPHOS) and for insulin gene in pancreatic islets.

19. FACTORII DE CRESTERE IGF1 SI IGF2 IN PATOLOGIA COPILULUI

Prof. Sorin Buzinschi
Universitatea Transilvania - Facultatea de Medicina Brasov

Factorii de crestere IGF formeaza un sistem care contine IGF1, IGF2, proteine transportoare (IGFBP) si receptori (IGF1r, IGH2r), cu rol in crestere si dezvoltare. Ei sunt implicati in stabilirea greutatii la nastere prin antagonismul IGF2 patern si IGF2r matern. Defectele genei IGF1 duc la nanismul Laron cunoscut pentru rezistenta la diabet si cancer. Expresia crescuta a genei IGF2 a fost implicata in aparitia sindromului Beckwith-Wiedeman, pe cand scaderea acesteia are ca rezultat sindromul Silver-Russell. Cresterea expunerii fetale la glucocorticoizi produce supresia genelor IGF1 si IGF2, scaderea dezvoltarii fetale si cresterea expresiei ARNm pentru receptorul IGF si IGFBP. Carentele nutritionale si in special de acid folic scad gradul de metilare al IGF2. Aceste modificari inductoare pentru diabet zaharat 2 si sindrom metabolic se mentin de-a lungul vietii, asa cum arata studii efectuate la mult timp de la initierea lor.

GROWTH FACTORS IGF1 AND IGF2 IN CHILDHOOD PATHOLOGY

Prof. Sorin Buzinschi
Transylvania University- Faculty of Medicine

Growth factors forms a system that contain IGF1, IGF2, binding proteins (IGFBP) and receptors (IGF1r , IGH2r) with role in growth and development. They are involved in determining birth weight by interposing paternal IGF2 and maternal IGF2r. IGF1 gene defects lead to Laron dwarfism known for resistance to diabetes and cancer. Increased expression of IGF2 gene has been implicated in Beckwith - Wiedemann syndrome , while its decrease results in Silver- Russell syndrome. Increased fetal exposure to glucocorticoids determines IGF1 and IGF2 genes suppression, decreased fetal development and increased expression of mRNA for IGF receptor and IGFBP. Nutritional deficiencies and especially folic acid decrease, lower the level of methylation of IGF 2. These inductive changes for type 2 diabetes and metabolic syndrome are maintained throughout life, as shown by studies conducted long after their initiation.

20. ACTUALITATI IN MANAGEMENTUL DIAGNOSTIC SI TERAPEUTIC AL DISTROFIEI MUSCULARE DUCHENNE

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Distrofia Musculară Duchenne (DMD), boala recesiv X linkata, ce afecteaza 1 la 3500 de nou nascuti de sex masculin, este cea mai freceventa si severa boala neuromusculara la om.

La nivel international, dezvoltarea terapiilor paleative şi a terapiilor genice, ofera alternativa intarzierii complicatiilor si cresterii sperantei de viata a persoanelor afectate de aceasta patologie.

Lucrarea de față isi propune validarea metodelor de diagnostic si terapie necesare pentru un management complet al DMD, care sa permita si pacientilor romani aceeasta perspectiva prognostica.

In studiu au fost inclusi 10 pacienti pentru care s-a aplicat un protocol diagnostic ce a cuprins elemente care au permis un diagnostic de certitudine: evaluare clinica, dozarea creatinfosfokinazei, examen imunohistochimic si analiza moleculara. Pentru toti pacientii diagnosticati a fost propus un plan de terapie ce a cuprins: corticoterapie, sedinte saptamanale de kinetoterapie, corectarea deformarilor osteo-articulare, sustinerea functiei cardiace.

Rezultatele obtinute dovedesc ca diagnosticul de certitudine are nevoie de explorari suplimentare: examenul imunohistochimic, esential in realizarea diagnosticului diferential si analiza moleculara ce stabileste diagnosticul si permite, în funcție de tipul mutației, includerea într-un studiu terapeutic țintit.

Terapiile bazate pe administrarea de corticosteroizi, suport repirator si cardiac, instituite precoce, cresc considerabil calitatea vietii pacientilor, dar si sansa de a fi inclusi in studiile clinice pentru diverse terapii aflate in desfasurare.

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ACTUALITIES IN THE MANAGEMENT OF THE DIAGNOSIS AND THERAPY OF DUCHENNE MUSCULAR DYSTROPHY

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Duchenne Muscular Dystrophy (DMD), X linked recessive inherited disease that affects 1 in 3500 male births, is the most frequent and severe human neuromuscular disease.

Palliative and gene therapy development offers the alternative of delaying the complications and increasing the life expectancy of the patient.The present work is trying to validate the necessary diagnosis and therapeutic methods for a complete management of Duchenne Muscle Dystrophy which would allow the same prognostic perspective for the Romanian patients.

The study was performed on 10 patients for which a decisional protocol comprising elements for a certainty diagnosis was applied: clinical evaluation, creatinphosphokinase evaluation, immunohistochemical examination and molecular analysis. A therapeutic plan was proposed for all the diagnosed patients: corticotherapy, weekly sessions of kinetotherapy, osteoarticular deformities correction, cardiac function sustainability.

The results prove that the certainty diagnosis needs further explorations: the immunohistological examination, essential for the differential diagnosis, as well as the molecular analysis which confirms the diagnosis and allows, according to the mutation type, the inclusion in a targeted therapeutic study (exon skipping).

Therapies based on corticosteroids administration, respiratory and cardiac support, established as soon as possible, significantly improve the patients` quality of life and also their chances of being included in ongoing internationally clinical trials for various therapies.

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21. PERSPECTIVELE DE CERCETARE SI DIAGNOSTIC IN CENTRUL DE MEDICINA GENOMICĂ, TIMIȘOARA

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Centrul de Medicina Genomica (CMG) se constituie ca un nucleu de cercetare și formare în cadrul Universității de Medicină și Farmacie "Victor Babeș" (UMFVBT). Dispunând de cele mai recente tehnologii și expertiză cheie pentru cercetarea OMICS, CMG va reuni colective prestigioase de cercetare, facând legatura cu cercetarea clinică și domeniile prioritare Orizont 2020.Principalele obiective ale Centrului de Medicină Genomică sunt: Cresterea capacitatii, calitatii si eficientei activitatii de cercetare, Dezvoltarea cunostintelor fundamentale, a cercetarilor experimentale si obtinerea de noi terapii innovative, Dezvoltarea resursei umane, Stimularea cercetarii in parteneriat si a transferului de tehnologie catre intreprinderi, Cresterea vizibilitatii internationale, dezvoltarea de noi colaborari.

Prin implementarea infrastructurii in cadrul CMG, se poate realiza un pol romanesc de cercetare in domeniul genomicii. Laboratoarele existente si modernizate tehnologic, precum si cele nou create, asigura translatarea cercetarii fundamentale dinspre si catre clinicieni. Obiectivul pe termen lung al CMG este de a imbunatati sistemul de sanatate printr-un management mai eficient al bolnavilor.Activitatile ce se vor desfasura in Centru vor fi legate de cercetarea in domeniul bolilor genetice si complexe. Preocuparile prioritare includ cercetarea bolilor genetice rare si descoperirea de noi perspective in tulburarile monogenice si aberatiile cromozomiale. Vor fi cercetate in paralel aspectele moleculare ale patologiei care asociaza componenta genetica cu factorii de mediu, studiul functiei mitocondriale si mecanismele epigenetice care stau la baza unor tulburari complexe.

Misiunea CMG este de a dezvolta și difuza instrumente care să permită cercetarea medicală de vârf, contribuind la descifrarea substratului molecular în domeniul bolilor genetice și complexe.

Cuvinte cheie: medicina genomica, boli genetice
Acest studiu a fost realizat in Centrul de Medicina Genomica din cadrul Universității de Medicină și Farmacie "Victor Babeș"Timisoara, Proiect POSCCE ID: 1854, cod SMIS:48749,„Centrul de Medicina Genomica *v2"*,
contract nr. 677/09.04.2015

PROSPECTS FOR RESEARCH AND DIAGNOSIS IN CENTER OF GENOMIC MEDICINE, TIMIŞOARA

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Center of Genomic Medicine (CMG) is conceived as a nucleus for research and training at the University of Medicine and Pharmacy "Victor Babes" (UMFVBT). Featuring the latest technology and key expertise for OMICS research, CMG will assemble a prestigious research collective, linking clinical research and priority areas of Horizon 2020.

The main objectives of the Center for Genomic Medicine are: increase the capacity, quality and efficiency of research, development of fundamental knowledge, experimental research, innovative therapies and human resource creation. Other

objectives include stimulating research in partnership and technology transfer to companies, growth of international visibility, develop new collaborations.

By implementing the infrastructure within CMG, the newly developed center will constitute a pole of Romanian research in genomics. Existing and technologically upgraded laboratories as well as newly created ones will ensure the translation of basic research from/to clinicians. CMG's long term objective is to improve the health care system through more effective patient management.

The activities to be developed in the center will be linked to research on genetic and complex diseases. Priority concerns include rare genetic disease research and discovery of new perspectives in monogenic disorders and chromosomal aberrations. In parallel, there will be investigations in the area of molecular aspects of the pathology that associate a genetic component with environmental factors, studies of mitochondrial function and epigenetic mechanisms that underlie some complex disorders.

CMG's mission is to develop and disseminate tools that enable leading medical research, helping to decipher the molecular substrate in complex genetic diseases.

Keywords: genomic medicine, genetic disorders

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HEMATOLOGIE ŞI ONCOLOGIE / HAEMATOLOGY AND ONCOLOGY

22. .RARITATI ÎN PATOLOGIA TUMORALĂ A PANCREASULUI NEUROENDOCRIN LA COPIL

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Tumorile pancreasului neuroendocrin sunt forme rare de afectare pancreatica la copil; ele se dezvolta din insulele Langerhans si se prezinta sub forma de tumori functionale sau nefunctionale. In cazul tumorilor functionale, simptomele sunt consecintele secretiei hormonilor specifici: insulin, glucagon, gastrina, peptidul intestinal vasoactive si altii. Aceste entitati pot fi asociate cu alte sindroame neoplazice cum ar fi MEN1, boala von Hippel Lindau sau neurofibromatoza. Insulinomul este caracterizat clinic prin triada Whipple (simptome de hipoglicemie, valori scazute ale glicemiei pe durata simptomatologiei si reversibilitatea simptomelor dupa administrarea de glucoza intravenos). Diagnosticul biochimic al insulinomului este stabilit de nivelul crescut al insulinemiei , peste 10 µU/mL (normal < 6 µU/mL), glicemia sub 40 mg/dl, peptidul C peste 2,5 ng/ml (normal <2 ng/ml), proinsulina mai mare de 25% din nivelul insulinei. Simptomele tumorilor functionale depind de tipul de hormon secretat, frecvent aparand diaree, scadere sau spor ponderal, eritem, vertij. Dezvoltarea imunhistochimiei a facilitat diagnosticarea tumorilor neuroendocrine pancreatice prin decelarea neuropeptidelor specifice. Chromogranin A este un marker cu o sensibiliate de pana la 70-85 %, dar valori crescute pot apare si in alte tipuri de neoplazii neuroendocrine (carcinomul de pulmon cu celule mici si unele tipuri de carcinoame de prostata). Rata de proliferare a tumorilor neuroendocrine este deasemenea evaluata cu ajutorul anticorpului Ki-67, acesta fiind acceptat de clasificarea OMS a acestor tumori . Pentru conduita terapeutica ulterioara este necesara identificarea tumorii primare cat si a eventualelor metastaze. Printre cele mai utilzate metode de diagnosticare si cele mai citate in literatura sunt ecografia abdominala, ecoendoscopia digestive, computer tomografia si imagistica prin rezonanta magnetica. Optiunile terapeutice sunt chirurgia curativa, paliativa, chimioterapia si terapia hormonala.

Lucrarea include si prezentarea unui pacient de sex masculin, in varsta de 11 ani, spitalizat in mai multe randuri pentru simptome neuroglicopenice, urmate de simptome hiperadrenergice. Initial a fost diagnosticat cu malnutritie si hipoglicemie (glicemie 38 mg/dl), ce s-a normalizat la administrarea de glucoza intravenos. Deasemenea copilul prezenta retard psihic moderat. In timpul simptomelor de neuroglicopenie, dupa o perioada de post, nivelul glicemiei era 41 mg/dl, iar valorile peptidului C si ale insulinei au fost crescute. Ecografia abdominala si computer tomografia au fost in limite normale, dar la examinarea prin rezonanta magnetica s-a descris o tumora la nivelul cozii pancreasului. Pe durata spitalizarii nivelul glicemiei a fost adesea in jurul a 50 mg/dl, iar nivelul hemoglobinei glicate de 4,12 %. Dupa excluderea altor cauze de hiperinsulinemism am considerat diagnosticul de insulinom. S-a intervenit chirurgical prin procedeul clasic, cu enucleerea tumorii de la nivelul cozii pancreasului, diagnosticul final de insulinom fiind stabilit de catre anatomopatolog. Originea

neuroendocrina a tumorii a fost stabilita de catre imunohistochimie pozitivă pentru cromogranin A, iar indexul de proliferare (agent nuclear KI-67) fiind de aproximativ 8%. Dupa interventia chirurgicala simptomatologia s-a remis.
Cuvinte cheie: tumorile pancreasului neuroendocrin, insulinomul, copil.

UNCOMMON PANCREATIC NEUROENDOCRINE TUMORS IN CHILDREN

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Pancreatic neuroendocrine tumors (PNTs) are uncommon pediatric diseases in children and arise from the pancreatic islets of Langerhans ; they present either as a functional or non-functional tumors. In functional tumors the symptoms are a result of the secretion of specific hormones such as insulin, glucagon, gastrin, and vasoactive intestinal peptide (VIP) or others. All these entities may be sporadic or associated with inherited neoplasia syndromes such as MEN1, von Hippel-Lindau disease or neurofibromatosis. *Insulinomas* are characterized clinically by the Whipple triad (Presence of symptoms of hypoglycemia, documented low blood sugar at the time symptoms and reversal of symptoms by glucose administration). The biochemical diagnosis of insulinoma is established during prolonged fasting (up to 72 h) when the following results are found: Serum insulin levels of 10 µU/mL or more (normal < 6 µU/mL); Glucose levels of less than 40 mg/dL; C-peptide levels exceeding 2.5 ng/mL (normal < 2 ng/mL); Proinsulin levels greater than 25% (or up to 90%) of immunoreactive insulin levels. Symptoms of the functional tumors depend of the type of secreted hormones, wich may cause diarrhea, weight loss, weight gain, skin rash, flushing or dizziness. The diagnosis is facilitated by the development of specific plasma or serum assays for peptides and amines produced by pancreatic neuroendocrine tumors as well as the development of immunohistochemistry. Chromogranin A is a sensitive (70–85%) marker of pancreatic neuroendocrine tumors, but nonspecific test because elevated levels may be find in other types of neuroendocrine neoplasms as well as small-cell lung, and even some prostate carcinomas. The rate of proliferation of a can be quantified by counting the percentage of cells that stain positive with the Ki-67 antibody. The Ki-67% has been widely accepted as the cardinal feature of tumor grading WHO NEN classification. The evaluation of the tumor extent and the identification of the exact site of the primary and metastatic lesions are necessary to decide whether a curative surgical approach is possible. Standard abdominal ultrasound, EUS, CT scan, and MRI study are used to assess tumor extent and the possible location of the primary lesion. Current treatment options for pancreatic neuroendocrine tumors are surgery, combination chemotherapy, hormone therapy.
The paper includes a synthetic presentation of a 11 years old boy who presented for several times with symptoms of neuroglycopenia with concurrent hyperadrenergic reaction. He was diagnosed with malnutrition associated hypoglycemia with low blood glucose level (38 mg/dL), which was normalized after intravenous glucose administration. Neurological examination detected mild mental. The fasting test was performed, which revealed inadequately high insulin and peptid C level with glucose level of 41 mg% and signs of neuroglycopenia. The image of pancreas was normal in the abdominal ultrasound and in IRM a tumor was found in the tail of pancreas. During hospitalization, multiple glucose level curve showed several values below 50 mg/dL associated with low glycated hemoglobin (4.12%). Urinary sample was negative for ketones. Taking into consideration the clinical findings and case history and excluding other causes of hyperinsulinemic hypoglycemia such as exogenous insulin administration and accidental ingestion of oral anti-diabetics, the case was finally interpreted as insulinoma. The patient underwent classic operation with enucleating the solitary tumor of the pancreatic tail and the the clinical diagnosis of insulinoma was confirmed by histopathology. The neuroendocrine origin of the tumor was confirmed using immunostaining for Chromogranin A associated to the neurosecretory granules in tumor cell cytoplasm and approximately 8 % of nuclear Ki67 positivity. Total remission of symptoms was achieved
Keywords: pancreatic neuroendocrine tumors, insulinoma, children.

23. IMPORTANŢA CLINICĂ A INVESTIGAŢIILOR BIOMOLECULARE IN HEMOFILIE

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In era medicinii moderne, a medicinii bazată pe evidenţe, explorarea bio-moleculară capată pentru diverse patologii o importanţă decisivă. Este şi cazul hemofiliei, o coagulopatie congenitală severă, la ora actuală perfect tratabilă.Investigarea substratului genic, responsabil de defecţiunea de sinteză a factorilor VIII/IX de coagulare permite pe lângă diagnosticul prenatal şi al purtătorilor tarei, sfatul genetic şi identificarea anomaliei responsabile, ce poate contribui la o abordare terapeutică personalizată.
Cu aceste premize, obiectivul nostru, propus în cadrul proiectului transfrontalier SCREENGEN a fost cel de identificare prin tehnica PCR la un număr de 73 bolnavi cu hemofilie, 24 de purtatoare a inversiei de intron 22 şi intron 1; la aceasta s-a adăugat cercetarea spectrului de anomalii genice la alţi 12 bolnavi, prin tehnica secvenţierii.

Rezultatele au relevat o frecvenţă ridicată (50.68% din bolnavii explorati) a inversiei de intron 22, recunoscut factor de predicţie pentru apariţia celei mai severe complicaţii, inhibitorii anti F VIII; în restul cazurilor s-au evidenţiat anomalii lipsite de semnificaţie prognostică deosebită: mutaţie missense şi non sense, inserţie sau deleţie mică pe un singur domeniu. Un indiciu valoros pentru atitudinea terapeutică l-a avut inversia de intron 22, care in 21,6 % din cazuri s-a insoţit de apariţia anticorpilor anti F VIII, obligând la schimbarea strategiei terapeutice.
In concluzie, bazaţi pe rezultatele noastre preliminare, considerăm că o cuprindere largă a tuturor bolnavilor cu hemofilie în acest screening bio-molecular ar fi o acţiune raţională şi cost-efectivă, propunându-ne-o pentru viitorul apropiat.
Cuvinte cheie: substrat bio-molecular, hemofilie, inversia intronului 22, defecte genice

CLINICAL IMPORTANCE OF BIO-MOLECULAR INVESTIGATIONS IN HEMOPHILIA

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In the era of modern evidence based medicine, bio-molecular exploration for various pathologies gains a decisive importance. It is also the case of hemophilia, a severe congenital coagulophaty, currently a treatable disorder. Gene substrate investigation, responsible for the altered synthesis of clotting factors VIII / IX, allows besides prenatal and carriers diagnosis, an identification of the responsible cause, contributing to a personalized therapeutic approach. With these assumptions, our objective, pursued in SCREENGEN project, was the identification, by PCR, in the DNA of 73 patients with haemophilia and 24 carriers, the inversion of intron 22 and intron 1; to this it was added the sequencing research of gene abnormalities in other 12 patients.
The results revealed a high frequency (50.68% of investigated persons) of intron 22 inversion, a recognized predictor for the occurrence of anti F VIII inhibitors (present in 21.6 % persons of our cohort); the other assessed mutations (missense and non-sense mutations, insertion or deletion in one small domain) lacked a predictive value..
In conclusion, based on our preliminary results, we believe that a broad inclusion of all patients with hemophilia in this bio-molecular screening would be a rational and cost-effective action in our country, intended for the near future.
Key words: bio-molecular substrate, haemophilia, intron 22 inversion, gene defect

HEPATOLOGIE / HEPATOLOGY

24. ROLUL MICROBIOTEI INTESTINALE IN PATOGENIA BOLILOR HEPATICE

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Microorganismele sunt prezente în număr de milioane de colonii în tractul gastrointestinal şi se cunoaşte rolul benefic al acestora asupra gazdei, iar modificările cantitative şi calitative sunt legate de alimentaţie, zona geografică, afecţiuni asociate, factori de mediu, tratament. Până de curând nu existau metode de identificare completă a microflorei bacteriene intestinale şi nici de întelegere a rolului compex al acesteia în starea de sănătate sau boală.
Datele recente de literatură sugerează că microbiota intestinală are rol cert în menţinerea homeostaziei. Modificările calitative (disbioza) sau cantitative (suprapopularea) ale microbiotei au rol dovedit în patogenia unor boli gastointestinale şi hepatice.
Autorii încearcă în această prezentare, folosind datele din literatură şi studiile experimentale publicate, să sintetizeze şi să ordoneze informaţiile referitoare la rolul patogenic al variaţiilor microbiotei în patogenia afecţiunilor hepatice precum: steatoza hepatică nonalcoolică asociată sau nu cu obezitate, steatohepatita, ciroza hepatică şi complicaţiile acesteia. Bolile hepatice au fost asociate de mult timp, ca mecanism fiziopatologic posibil, cu afectarea cantitativă sau calitativă a microbiotei. Factorii extrinsici pot determina disbioză, având ca
rezultat inflamaţie intestinală, modificarea funcţiei de bariera a tractului digestiv, translocarea produşilor bacterieni ce pot produce injurie şi inflamaţie hepatică. Metaboliţii microbieni prezenţi într-un tract intestinal cu disbioză si factorii legaţi de gazdă sunt egal implicaţi în producerea şi progresiunea bolii hepatice.
Scopul studiilor este de a dovedi eficienţa utilizării probioticelor în terapia steatozei hepatice, chiar ca mijloc terapeutic de urgenţă în cazurile de steatohepatită sau ciroză hepatică.

THE ROLE OF GUT MICROBIOTA IN THE PATHOGENESIS OF LIVER DISEASES

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Microorganisms are present in number of million colonies in the gastrointestinal tract and it is known their beneficial role on the health of the host, and the quantitative and qualitative changes depend on nutrition, geographic and environmental factors, associated diseases, treatment. Until recently there were no methods for enterely identification of intestinal microflora and its compex role in health or disease was lnor completly understood.

Recent literature data suggest that gut microbiota has a certain role in maintaining homeostasis.

Qualitative changes (dysbiosis) or quantitative (overgrowth) of microbiota have a proven role in the pathogenesis of the gastrointestinal and liver diseases.

The authors tried in this presentation, using literature data and published experimental studies, to synthesize and rank information about the pathogenic role of microbiota in the pathogenesis of the liver diseases such as hepatic steatosis associated or not with obesity, steatohepatitis, liver cirrhosis and its complications. Liver diseases were associated for long time, as pathophysiological possible mechanism, with the quantitive or qualitative variability of the microbiota. Extrinsic factors may cause dysbiosis, resulting in intestinal inflammation, change of the digestive tract barrier function, bacterial products translocation that may cause liver injury and inflammation. Microbial metabolites present in a intestinal tract with dysbiosis and host-related factors are equally involved in producing and progression of liver disease.

The purpose of the studies is to prove the efficiency of using probiotics in therapy of hepatic steatosis, even as a therapeutic tool in cases of emergency steatohepatitis and cirrhosis.

25. ABORDAREA PRACTICA A BOLII FICATULUI GRAS NON-ALCOOLIC LA COPIL SI ADOLESCENTI

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Boala ficatului gras non-alcoolic(BFGNA) este o conditie multifactoriala responsabila de dereglarea sistemica a lipidelor, care la nivel hepatic determina de la steatoza la steatohepatita non-alcoolica(NASH), ciroza si carcinom hepatocelular.

Prevalenta BFGNA si a obezitatii a inregistrat cresteri semnificative, devenind cea mai frecventa cauza a bolii hepatice cronice la copii si adolescent,fiind de 3-10% in America de Nord si Sud, Europa, Asia, Australia si pina la 80% la copiii obezi din tarile industrializate.

Patogeneza BFGNA este partial cunoscuta, acceptandu-se implicarea deopotriva a factorilor genetici si de mediu. Intelegerea mai complexa a mecanismelor patogenetice contribuie la dezvoltarea instrumentelor diagnostice non-invazive si la identificarea de noi terapii farmacologice.

O problema persistenta in diagnosticul si monitorizarea BFGNA este lipsa unei metode simple si accesibile, care sa inlocuiasca biopsia hepatica. Un algoritm practic are ca prima linie efectuarea ALT si a ecografiei hepatice la toti copiii peste 3 ani, supraponderali si obezi. Daca unul din aceste teste sugereaza ficatul gras se verifica rezistenta la insulina. Daca exista rezistenta la insulina se suplimenteaza explorarea cu biomarkeri ai inflamatiei si fibrozei, sugestivi pentru NASH(citokeratina-18; Panoul European de Fibroza(ELF)=acid hyaluronic, colagen III si inhibitorul tisular al metaloproteinazei 1). Sunt in evaluare si alti biomarkeri si metode imagistice(RMN, Fibroscan, ARFI). Totusi, biopsia hepatica ramane standardul de aur in diagnosticarea si stadializarea BFGNA la copii si adolescenti.

Terapia de prima linie se axeaza pe modificarea alimentatiei si a stilului de viata, printr-o abordare multidisciplinara. Lipsa de raspuns sau non-complianta la masurile initiale lasa loc terapiilor farmacologice(Vitamina E, Metformin, probiotice, Acid ursodeoxicolic, Acizi grasi polinesaturati).

Nu exista un consens general privind eficacitatea acestor medicamente in BFGNA pediatrica. Sunt necesare studii adecvate si controlate inainte de recomandarea lor universala.

PRACTICAL APPROACH TO NON-ALCOHOLIC FATTY LIVER DISEASE IN CHILDREN AND ADOLESCENTS

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Non-alcoholic fatty liver disease (NAFLD) is a multifactorial condition, responsible for a systemic disorder of lipid, which can cause in the liver from steatosis to non-alcoholic steatohepatitis (NASH), cirrhosis and hepatocellular carcinoma.

The prevalence of NAFLD and obesity recorded significant increases, becoming the most common cause of chronic liver disease in children and adolescents, being 3-10% in North and South America, Europe, Asia, Australia and up to 80% of obese children in industrialized countries.

The pathogenesis of NAFLD is partially known, we accept involve of both genetic and environmental factors. A complex understanding of the pathogenetic mechanisms may contribute to the development of non-invasive diagnostic tools and to identify new pharmacological therapies.
A persistent problem in the diagnosis and monitoring of NAFLD is the lack of simple and affordable methods, witch can replace liver biopsy. A practical algorithm has, as the first line, detection of ALT and liver ultrasonography, in all children over 3 years, overweight and obese.
If any of these tests suggest fatty liver, insulin resistance is checked. If insulin resistance exist, the exploration is supplemented with inflammation and fibrosis biomarkers, suggestive for NASH (cytokeratin-18, European Panel of Fibrosis (EPF) = hyaluronic acid, collagen III and tissue inhibitor metalloproteinase 1).
There are in evaluation other biomarkers and imaging methods (MRI, Fibroscan, ARFI). However, liver biopsy remains the gold standard in the diagnosis and staging of NAFLD in children and adolescents.
First-line therapy focuses on changing eating and lifestyle, through a multidisciplinary approach. Lack of response or non-compliance to the initial measures leave room for pharmacological therapies (Vitamin E, Metformin, probiotics, Ursodeoxycholic Acid, Polyunsaturated Fatty Acids).
There is not a general consensus regarding the efficacy of these drugs in pediatric NAFLD. Adequate and controlled studies are needed, before their universal recommendation.

26. HEPATOPATIA ASOCIATA FIBROZEI CHISTICE (MUCOVISCIDOZA)

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Introducere:Fibroza chistica (FC) este cea mai frecventă boală genetică monogenică, cu transmitere autozomal recesiva, caracterizată printr-o polimorfism clinic impresionant și cu o evolutie nefastă. Hepatopatia asociata FC a doua cauza non-pulmonară de deces la pacientii cu FC, care, o dată cu creșterea speranței de viață, a devenit o problemă importantă de management. Factorii predispozanti, cum ar fi sexul masculin, insuficiență pancreatică, ileusul meconial și mutațiile severe sunt incriminate de a influența apariția fibrozei asociate bolii hepatice chistica (HFC). Material si metode: Studiul nostru a inclus 174 de pacienti cu FC, monitorizati în Centrul Național de Fibroza Chistică, Timișoara, România. Acestia au fost monitorizati astfel, la fiecare vizită de rutină, s-a practicat examenul clinic, teste biochimice hepatice , ultrasonografie hepatica și alte metode, cum ar fi elastografia tranzitorie, biopsie, în cazuri selectate. Rezultate: Șaizeci și șase de pacienți, cu vârsta medie la diagnostic 4.33 ani, au fost diagnosticat cu HFC, fără diferență semnificativă între femei și bărbați. HFC a fost frecventă la pacienții cu vârsta de peste opt ani, cu istoric de ileus meconial, purtatori de mutatii severe (p = 0,002). Insuficiență pancreatică, deși prezentă în 75% din pacienții cu HFC nu a fost confirmat ca factor de risc, nici genul masculine, în studiul nostru. Concluzie: Copiii cu FC mai mari de opt ani, purtatori ai unui genotip sever, cu istoric pozitiv de ileu smeconial, sunt mai predispusi la HFC.

CYSTIC FIBROSIS ASSOCIATED LIVER DISEASE(CFLD)

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Background:Cystic fibrosis (CF) is the most frequent monogenic genetic disease, autosomal recessive transmitted, characterized by an impressive clinical polymorphism and appreciative fatal prospective. Liver disease is the second non-pulmonary cause of death in cystic fibrosis, which, with increasing life expectancy, became an important management problem. Predisposing factors like male gender, pancreatic insufficiency, meconium ileus and severe mutation are incriminated to influence the occurrence of cystic fibrosis associated liver disease (CFLD). Methods:Our study included 174 patients with CF, monitored in the National Cystic Fibrosis Centre, Timisoara, Romania. They were routinely followed-up by clinical assessment, liver biochemical tests, ultrasound examinations and other methods like transient elastography, biopsy, in selected cases. Results:Sixty-six patients, with median age at diagnosis 4.33 years, diagnosed with CFLD, without significant gender gap. CFLD was frequent in patients aged over eight years, with meconium ileus history, carriers of severe mutations (p=0.002). Pancreatic insufficiency, although present in 75% of patients with CFLD was not confirmed as risk factor, not male gender, in our study. Conclusion:CF children older than eight years, carriers of a severe genotype, with a positive history of meconium ileus, were more likely predisposed to CFLD.

27. . FACTORI DE PROGNOSTIC ÎN EVOLUŢIA HEPATOPATIILOR ACUTE ŞI CRONICE LA COPIL

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Proiect Parteneriate PN-II-PT-PCCA-2011-3.2-0917

Patologia hepatică la copil este variată, cu manifestări hepatice variate de la creşterea uşoară a transaminazelor decelată întâmplător până la insuficienţa hepatică fulminantă. Evoluţia bolii poate fi foarte severă, cu deces în lipsa unor măsuri eficiente luate la timp. Nivelul transaminazelor nu reprezintă un factor care să reflecte gravitatea bolii şi să determine prognosticul în patologia hepatică în general.
În patologia colestatică neonatală atrezia biliară ocupă un loc important prin evoluţia severă spre ciroza şi deces în lipsa transplantului hepatic. Printre cei mai importanţi factori de pronostic sunt diagnosticul precoce, vârsta la care se efectuează hepatoportoenterostomia, gradul de specializare al centrului respectiv în această tehnică.
Considerată iniţial ca ireversibilă, fibroza hepatică este un proces dinamic, cu potenţial de reversibilitate. Identificarea rapidă, evaluarea corectă şi intervenţia terapeutică în timp util ar putea limita consecinţele nefavorabile ale cirozei hepatice. Biopsia hepatică este standardul în evaluarea fibrozei hepatice, dar în unele situaţii valoarea acesteia este pusă la îndoială datorită limitelor tehnice şi a riscurilor. Evaluarea neinvazivă a fibrozei hepatice se bazează pe determinarea serică a unor markeri implicaţi în fibroză sau pe metode imagistice şi tinde să înlocuiască biopsia hepatică.
Pentru aprecierea severităţii cirozei hepatice exista scoruri de pronostic. Unele din aceste scoruri, cum e cazul PELD, sunt utilizate pentru a evalua necesitatea efectuării transplantului hepatic şi urgenţa acestuia.
În cazul insuficienţei hepatice acute s-au imaginat mai multe astfel de scoruri, care includ nivelul transaminazelor, factorii coagulării, albumina. În boala Wilson cu evoluţie fulminantă un astfel de scor este utilizat pentru prognosticul evoluţiei şi necesitatea transplantului.
În concluzie, este importantă cunoaşterea factorilor care influenţează prognosticul şi evoluţia acestor pacienţi pentru a stabili nevoia transferului într-un centru specializat în hepatologie pediatrică sau chiar a transplantului hepatic.
Cuvinte cheie: scor prognostic, atrezie biliară, insuficienţă hepatică, fibroză, ciroză, boala Wilson, transplant hepatic

PROGNOSTIC FACTORS IN ACUTE AND CHRONIC LIVER DISEASES IN CHILDREN

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Partnership Project PN-II-PT-PCCA-2011-3.2-0917

Liver diseases in children have variable clinical manifestations, from asymptomatic increase of transaminases to fulminant liver failure. The disease evolution could be very severe, even fatal without right treatment on right moment. The level of transaminases is not a very valuable for predicting the severity of the disease.
In neonatal cholestasis, biliary atresia is very important as it could have a severe evolution to cirrhosis and death. The most important prognostic factors in biliary atresia are the precocity of the diagnostic, the age at the surgical intervention, the experience of the surgical team.
In acute liver failure there are many prognostic scores that include level of transaminases, coagulation factors or albumin level, with some differences depending on the etiologies. One of the clinical onset forms in Wilson disease is the fulminant liver failure. The severity and the need of liver transplantation could be assessed using a dedicated prognostic score.
Even considered irreversible, liver fibrosis is a dynamic process potential reversible. The quick diagnostic, correct evaluation and timely therapy could limit the unfavorable consequences of liver cirrhosis. Liver biopsy is the gold standard in fibrosis evaluation, but sometimes the risks could limit its value. Noninvasive evaluation of liver fibrosis is based on measuring the serum level of makers involved in fibrosis or on imagistic methods and tend to replace the liver biopsy.
In liver cirrhosis, there are different scores used for evaluation of the severity. Among those scores, PELD (pediatric end-stage liver disease) is used to evaluate the need of liver transplantation and also the urge of it.
In conclusion, knowing the prognostic factors in different liver diseases is very important in order to assess the need of high specialized care in a liver center including liver transplantation in order to increase the survival of those children.
Keywords: prognostic scores, biliary atresia, liver failure, fibrosis, cirrhosis, Wilson disease, liver transplantation

IMAGISTICA / MEDICAL IMAGING

28. LOCALIZARI ABDOMINALE ALE LIMFOAMELOR PEDIATRICE – CE TREBUIE SA STIM

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Patologiile canceroase sunt rare in pediatrie. Limfoamele Hodgkin si cele non hodgkiniene ocupa locul trei dupa leucemii si tumorile cerebrale, reprezentind aproximativ 10 % din cancerele pediatrice. LNH reprezinta aproximativ 57 %

dintre limfoamele la copil si MDH aproximativ 43 %. Cele doua entitati sunt boli ale sistemului limfocitar dar MD are o evolutie progresiva cu simptomatologie ganglionara pe primul plan pe cand LNH au o prezentare zgomotoasa in majoritatea cazurilor abdominala dar si toracica sau ORL , cu evolutie rapida angajand prognosticul vital , si necesitant un tratament in urgenta. Diagnosticul de certitudine necesita o analiza histologica si imunohistochimica al celulelor tumorale dupa prevelare (biopsie etc) . Diversele metode imagistice au un rol fundamental in stabilirea diagnosticului , realizarea bilantul de extensie , eliminarea altor diagnostice posibile , evaluarea in cursul tratamentului. Lucrarea de fata isi propune ca prin cazuri concrete sa faca o trecere in revista a localizarilor abdominale ale limfoamelor la copil si adolescent punand accent pe rolul imagisticii in diagnosticul, extensia si urmarirea in timpul tratamentului.

ABDOMINAL LOCALIZATION OF PEDIATRIC LYMPHOMAS- WHAT DO WE HAVE TO KNOW?

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Background: Cancer is rare in pediatric pathology. Non Hodgkin and Hodgkin lymphoma are on the third place after leukemia and brain tumors, representing approximately 10% of pediatric cancers. NHL lymphomas represent approximately 57% of lymphomas in children and MDH about 43% .The two entities are diseases of the lymphocytes but is developing MD progressive symptomatic first NHL when they plan a presentation noisy but in most cases the abdominal and thoracic and ENT rapidly evolving employing vital prognosis and required treatment in an emergency. Correct diagnosis requires histological and immunohistochemical analysis of tumor cells after prelevation (biopsy etc).
The various imaging methods play a key role in diagnosis, achieving balance extension, eliminating other possible diagnoses, assessment during treatment.
The aims of the paper is to make a review of abdominal lymphoma localization in children and adolescents focusing on the role of imaging in the diagnosis, extension and monitoring during treatment.

29. ROLUL ULTRASONOGRAFIEI PULMONARE IN PATOLOGIA PEDIATRICA

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Introducere: Ultrasonografia este o metoda larg utilizata in diagnosticul diverselor patologii; ultrasonografia pulmonara aduce date immediate asupra patologiei pulmonare, fiind extrem de utila in evaluarea patologiei acute ca pneumotorax, pneumonie, edem pulmonar, pleurezie.
Material si metoda: Lucrarea de fata isi propune sa prezinte rolul pe care il poate avea ultrasonografia pulmonara(USP) in evaluarea patologiei respiratorii la copil. Rezultate: Lucrarea prezinta o serie de cazuri de pneumonie, pleurezie, tumori si malformatii pulmonare, frecvent intalnite in patologia pediatrica. Pentru depistarea colectiilor pleurale mici , US pulmonara are o sensibilitate mai buna decat radiografia, diagnosticul consolidarilor pulmonare se poate face rapid precum si monitorizarea neiradianta a evolutiei focarului pneumonic. In ceea ce priveste patologia cronica, ca astmul bronsic, bronhopneumopatia cronica, patologia tumorala pulmonara, rolul ecografiei este relativ limitat, atat de dimensiunile si structura acestora leziunilor cat si de de poate fi evidentiata de asemenea, ramanand mai dificil de evaluat si patologia cronica de tip bronsiectatic.
Concluzii: Ultrasonografia pulmonara este o metoda utila in evaluarea si monitorizarea anumitor patologii frecvent intalnite la copil.

LUNG ULTRASOUND IN PEDIATRIC PATHOLOGY

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Introduction: Ultrasound is a method widely used in medical diagnosis; pulmonary ultrasonography add immediate data on divers pulmonary pathology, being extremely useful in the evaluation of acute pathology like pneumothorax, pneumonia, pulmonary edema, pleural effusion.
Methods: This paper aims to present the role of lung ultrasonography (USP) in evaluating respiratory pathology in children.
Results: This paper presents a number of cases of pneumonia, pleurisy, lung tumors and malformations, commonly found in pediatric pathology. Lung US has a superior sensitivity than radiography in detection of small pleural effusion; diagnosis of pulmonary consolidations can be done quickly also the non-irradiating monitoring of the pneumonia evolution. In terms of chronic pathology as asthma, chronic pulmonary, lung tumor, ultrasound has a relatively limited role, depending on their size and structure of the lesion, also remains difficult to assess and chronic bronchiectasis.
Conclusions: Pulmonary ultrasound is a useful method in the evaluation and monitoring of certain diseases commonly found in children.

30. UTILIZAREA UROSONOGRAFIEI CU SUBSTANȚĂ DE CONTRAST ÎN DETECTAREA REFLUXULUI VEZICOURETERAL LA COPIL

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Urosonografia cu substanță de contrast a fost dezvoltată mult în ultima decadă în pediatrie, această metodă dovedindu-se o alternativă eficientă în decelarea refluxului vezicoureteral, avantajul major constând în lipsa de iradiere. Metoda presupune introducerea prin cateter intravezical a substanței de contrast (Levovist sau Sonovue), în combinație cu ser fiziologic. Ulterior, folosind softul de contrast al ecografului, se urmărește în timp real, ascensionarea substanței de contrast în tractul urinar superior, prezentă în cazul refluxului. În Clinica Pediatrie III, am introdus această metodă de diagnostic de aproximativ 1 an, folosind ca agent de contrast Sonovue. O parte din cazuistica abordată, este prezentată în această lucrare, prezentând un scurt istoric al fiecărui pacient, din care reiese suspiciunea de diagnosic și exemplificând la fiecare caz cu imaginile obținute. Acestea relevă o dată în plus acuratețea acestei metode în diagnosticul refluxului vezicoureteral și rezultatele ne stimulează în continuarea aplicării tehnicii și a perfecționării acesteia în domeniul urologiei pediatrice.

VOIDING UROSONOGRAPHY WITH ULTRASOUND CONTRAST AGENTS FOR DETECTION OF VESICOURETERAL REFLUX IN CHILDREN

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Voiding urosonography with contrast agents has been developed over the past decade in pediatrics, while this method proved to be an effective alternative to detect vesicoureteral reflux, the major advantage consisting in the absence of irradiation. This method involves intravesical injection, by urinary catheter of the contrast agent (Levovist or SONOVUE) in combination with normal saline. Subsequently, using the contrast software, real time, we could detect the ascension of the contrast in the upper urinary tract, present in the case of reflux. In Pediatrics III, we introduced this method of diagnosis about 1 year using SonoVue as a contrast agent. A part of the cases addressed, is presented in this paper, presenting a brief history of each patient, which shows suspicion of diagnostic and exemplifying each case with the images. This images reveals once again the accuracy of this method in the diagnosis of vesicoureteral reflux and results encourages us to continue improving the technology and its application in the field of pediatric urology.

31. APORTUL ULTRASONOGRAFIEI POSTNATALE IN EVALUAREA HIDRONEFROZELOR CONGENITALE

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Hidronefroza congenitală este cea mai frecventă anomalie renourinară, decelată ecografic antenatal sau imediat postnatal. În majoritatea situațiilor, este vorba de o hidronefroză de grad mic, ce remite spontan în evoluție. O problemă controversată încă, este atitudinea optimă în cazul hidronefrozelor congenitale severe, respectiv oportunitatea inițierii în afara ultrasonografiei (US) si a altor investigații imagistice, precum și stabilirea indicației și momentul intervenției chirurgicale. Rolul US în acest algoritm decizional se regăsește în monitorizarea obiectivă a dilatării căilor urinare si a grosimii parenchimului. În acest sens, măsurăm o serie de parametri reproductibili (diametrul anteroposterior și aria pelvisului, tije caliceale, grosimea parenchimului). Se va prezenta o serie de cazuri de hidronefroză severă, monitorizate în clinica noastră, în care US a avut un rol important în orientarea diagnosticului și a altor investigații necesare la fiecare caz în parte. Indicația chirurgicală a fost luată prin decizie comună (pediatru, urolog, radiolog, specialist medicină nucleară). În majoritatea cazurilor, însă, printr-o monitorizare ecografică atentă, s-au temporizat sau chiar evitat alte investigații, întrucât s-a dovedit în timp o evoluție spontan favorabilă a hidronefrozei. În concluzie, US postnatală este investigația imagistică indispensabilă în conduita terapeutică modernă a hidronefrozelor congenitale.

THE CONTRIBUTION OF KIDNEY ULTRASONOGRAPHY IN THE MANAGEMENT OF ANTENATAL DETECTED HYDRONEPHROSIS

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Congenital hydronephrosis is the most common reno-urinary anomaly, detected on antenatal ultrasound or immediately after birth. In most situations, it is a low-grade hydronephrosis, which resolves spontaneously in time. A controversial issue still is the best attitude in severe hydronephrosis, regarding when it is appropriate to initiate, outside ultrasonography (US), other imaging investigations and determining the indication and timing of surgery. US role in this decision algorithm is found in the objective monitoring of urinary tract dilatation. In this regard, reproducible parameters are measured (anteroposterior diameter and pelvic area, the calyx dilatation, thickness of renal parenchyma). We present a series

of cases of severe hydronephrosis, monitored in our clinic, in which the US has played an important role in guiding diagnosis and other investigations needed in every case. Surgical indication was made by common decision (pediatrician, urologist, radiologist, nuclear medicine specialist). In most cases, however, through a careful ultrasonographic monitoring, further investigations were delayed or even avoided, as time proved favorable spontaneous evolution of hydronephrosis. In conclusion, postnatal US investigation is indispensable in modern management of congenital hydroneprosis.

IMUNOLOGIE / IMMUNOLOGY

32. HIPOGAMAGLOBULINEMIA – SEMN MAJOR IN DIAGNOSTICUL IMUNODEFICIENTELOR PRIMARE

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Hipogamaglobulinemia este definita prin scaderea nivelului seric al gamaglobulinelor sub 2 deviatii standard fata de valorile normale pentru varsta si poate fi intilnita la toate varstele. Dupa excluderea cauzelor secundare (sindrom nefrotic, enteropatie cronica, limfom, timom, medicamente), hipogamaglobulinemia ramane expresia majora a imunodeficientelor umorale sau cu componenta umorala. Acestea includ cele mai frecvente imunodeficiente primare, reprezentand aproximativ 50% din totalul lor. Mecanismele de producere sunt complexe si multiple, incepand de la defecte ale maturarii limfocitului B (principalul actor) situate precoce (boala Bruton, unele imunodeficiente combinate severe) sau terminal (imunodeficienta comuna variabila), defecte ale cooperarii dintre limfocitul B si T (unele imunodeficiente combinate severe) , defecte ale switch-ului pentru sinteza de imunoglobuline G (sindroamele de hiperIgM) sau defecte intrinseci ale limfocitului B. Expresia clinica esentiala consta in infectii recurente, persistente in ciuda tratamentului corect, de severitate neobisnuita cu germeni incapsulati dar si in boli autoimune si cancer, cu un risc mai mare decat pentru populatia generala. Explorarile necesare stabilirii tipului de imunodeficienta sunt multiple si sofisticate cuprinzand imunograma, subclasele de IgG, subpopulatiile limfocitare, raspunsul la vaccinare si chiar teste genetice, obligatorii pentru diagnosticul definitiv in anumite tipuri de imunodeficiente primare. Tratamentul se adreseaza, pe de o parte complicatiilor infectioase, autoimune, maligne iar pe de alta parte, in functie de tipul de imunodeficienta, profilaxiei acestora prin administrarea periodica de imunoglobulina sau rezolvarii defectului imun prin transplant medular. Un diagnostic precoce si precis si un tratament corect vor oferi acestor bolnavi speranta la o viata normala.

HYPOGAMMAGLOBULINEMIA – MAJOR SIGN IN PRIMARY IMMUNODEFICIENCIES DIAGNOSIS

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Hypogammaglobulinemia is defined by decreased gammaglobulins serum level less than 2 SD for age specific normal values and it can be found at any age. After exclusion of secondary causes (nephrotic syndrome, chronic enteropathy, lymphoma, thymoma, drugs), hypogammaglobulinemia represents the major expression for humoral immunodeficiencies or for those with humoral component. These include the most frequent primary immunodeficiencies, being approximately 50% from all of them. The mechanisms are complex and diverse represented by B lymphocyte (main actor) maturation defects, in early stage (Bruton disease, some severe combined immunodeficiencies) or in terminal stage (common variable immunodeficiency), defects in between B and T cell cooperation (some severe combined immunodeficiencies), immunoglobulin class switch recombination defects (HyperIgM syndrome) or intrinsic B cell defects. The clinical picture consists of infections with encapsulated germs: recurrent, persistent despite the correct treatment, with an unusual severity , but also of autoimmune disorders and cancer, with a higher risk than in general population. Laboratory tests required for the type of immunodeficiency diagnosis are multiple and sophisticate, including immunogram, IgG subclasses, lymphocyte subpopulations, response to vaccination and even genetic testing obligatory for definitive diagnosis in some primary immunodeficiencies. On one hand, the treatment addresses to infectious, autoimmune, malignant complications and on the other hand, in function of the type of immunodeficiency, for infections prevention using regular immunoglobulin administration or for cure of the immune defect trough bone marrow transplantation. An earlier and accurate diagnosis and a correct treatment offer to these patients the chance for a normal life.

33. IMUNODEFICIENTELE PRIMARE – CENUSAREASA IMUNOLOGIEI

Margit Serban
Academia Romana de Stiinte Medicale

Imunodeficientele primare (IDP), desi consecinta unor defecte ale sistemului imun, sunt dovada complexitatii si diversitatii acestuia. Cu o incidenta de 1:500 persoane, IDP nu mai sunt considerate boli rare, insa fiecare din cele peste 250 de tipuri de IDP ramane o boala rara. Sunt boli orfane datorita faptului ca sunt necunoscute de catre medici, aproximativ 80% dintre pacienti fiind nediagnosticati, incorect diagnosticati sau diagnosticati tardiv. Diagnosticul precoce este esential in IDP si chiar salvator de viata in imunodeficientele combinate severe, motiv pentru care in tot mai multe tari se discuta introducerea screeningului neonatal pentru acestea. Cel mai frecvent semn sugestiv de IDP ramane susceptibilitatea patologica la infectii cu anumite tipuri de agenti patogeni, cu localizare si de severitate neobisnuita, recurente si persistente in ciuda tratamentului corect efectuat. Falimentul cresterii si istoricul familial pozitiv sau de deces prin infectie la varsta mica sunt de asemenea foarte sugestive pentru IDP.O alta categorie importanta de manifestari care pot indica o IDP sunt cele de dereglare imuna: granuloamele, bolile autoimune, febra periodica, eczema, limfoproliferarea si diareea cronica neexplicata. Diagnosticul se face etapizat impunand investigatii sofisticate si costisitoare de care dispun numai centrele specializate in IDP. Tratamentul include substitutie cu imunoglobuline, citokine, profilaxia infectiilor, transplantul medular si terapia genica. Diagnosticul precoce ramane cheia succesului in IDP, aceasta insemnand mai putine sau absenta complicatiilor, doze mai mici de imunoglobulina, o calitate si o durata a vietii mai mari si o sansa mai mare de vindecare in IDP combinate severe.

PRIMARY IMMUNODEFICIENCIES – THE CINDERELLA OF IMMUNOLOGY

Margit Serban
Romanian Academy of Medical Sciences

Primary immunodeficiencies (PID) are the result of immune system's defects and are the evidence of its complexity and diversity. With an incidence of 1:500 persons, PID are no longer rare disorders, but each of the more than 250 types of PID is still a rare disease. They are also orphan disorders because they are ignored by doctors, approximately 80% of patients being undiagnosed, misdiagnosed or late diagnosed. An earlier diagnosis is essential in PID and often is saving the life such in severe combined immunodeficiencies, reason of which, in a lot of countries there is taken in discussion the opportunity of neonatal screening. The most suggestive sign for PID is the pathological susceptibility for infections with certain germs, sometimes opportunistic, unusual site and severity, recurrent and persistent despite the correct treatment. Failure to thrive and positive family history or death due to infection in infancy are also very suggestive for PID. Another important category of clinical manifestations indicating a PID consist of immune dysregulation signs: granuloma, autoimmunity, periodic fever, irregular eczema, lymphoprolipheration and unexplained chronic diarrhea. Diagnosis is performed step by step, using sophisticated and expensive explorations possible to be performed only in PID specialized centers. The treatment includes immunoglobulin substitution, cytokine, infections prevention, bone marrow transplantation and gene therapy. An early diagnosis remains the key of success in PID with less or absent complications, smaller doses of immunoglobulin, a higher quality and expectancy of life and a better chance for curing in severe combined PID.

34. ARPID – INTRE SPERANTE SI REALIZARI

Artemiza Baldea
Asociatia Romana a Pacientilor cu Imunodeficiente Primare (ARPID)

Constituirea asociatiei de pacienti cu imunodeficiente primare (IDP) reprezinta nu numai un mijloc prin care acesti bolnavi comunica dar mai ales o modalitate de lupta pentru un acces mai bun la diagnostic si tratament alaturi de mica echipa de medici din Romania implicata in managementul IDP. ARPID s-a constituit in 2010 la initiativa unui medic si a catorva pacienti adulti si parinti si numara astazi peste 60 de membri. In 2011 ARPID devine membru al asociatiei internationale pentru pacienti cu IDP, IPOPI. Intrucat pacientii adulti cu IDP umorale din tara noastra nu aveau acces la tratamentul cu imunoglobulina, in 2011 impreuna cu medici a fost depus la Ministerul Sanatatii un memoriu in acest sens, reusind introducerea unui program destinat adultilor cu IDP. In 2014 ARPID organizeaza primul meeting national al pacientilor cu IDP. Ca urmare a faptului ca in tara noastra 80% dintre bolnavii cu IDP raman nediagnosticati sau sunt gresit diagnosticati, aceasta asociatie a luptat pentru constientizarea populatiei si a medicilor in ceea ce priveste IDP, fiind alaturi de centrele medicale consacrate. Acest obiectiv s-a realizat prin publicarea de brosuri, emisiuni radio si TV, actiuni de binefacere in sprijinul societatii, implicarea elevilor din scoli si, nu in ultimul rand, prin participarea la o masa rotunda in Parlamentul European. O mare realizare o consideram organizarea a doua editii de tabere pentru copiii cu IDP, ocazie cu care acestia se pot intalni, isi pot impartasi nevoile, sperantele, temerile si se bucura de o vacanta minunata la munte.

ARPID – BETWEEN HOPES AND ACHIEVEMENTS

Artemiza Baldea
Romanian Association for patients with primary immunodeficiencies (ARPID)

The establishing of association for patients with primary immunodeficiencies (PID) represents not only a communication way but also a modality of fighting for a better access to diagnosis and treatment, together with the small Romanian doctors team engaged in PID management. ARPID constituted in 2010 at the initiative of a doctor and several parents and adults patients and today it counts more than 60 members. In 2011 ARPID became member of international PID patients organization, IPOPI. Because adult patients with humoral PID from our country didn't have access to immunoglobulin treatment, in 2011 together with some doctors we submitted a memoir to the Health Ministry, succeeding to introduce a program for PID adults. In 2014 ARPID organized the first national PID patients meeting. Taking in account that in our country, 80% from PID patients are not diagnosed or are misdiagnosed, this association promotes PID awareness together with medical centers. This objective is achieved trough brochures publishing, radio and TV broadcast, students involvement, and not at last, participation to a round table on this issue in the European Parliament. A great achievement we consider the organization of two summer camps for PID children where they meet each other and share their needs, hopes, fears but also they enjoy a wonderful mountain vacation.

NEFROLOGIE / NEPHROLOGY

35. PERSPECTIVE SI STRATEGII TERAPEUTICE NOI IN STATUSUL PROTEINURIC CRONIC LA COPIL

BALGRADEAN MIHAELA
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Proteinuria este o problema diagnostica in patologia pediatrica. Persistenta proteinuriei, de nivel nefrotic sau nu, reprezinta un semn sever de boala renala, fiind un marker al bolii renale cronice (BRC) atat la adult cat si la copil. La copil, orice afectare renala ce poate determina in timp glomeruloscleroza (infectii severe, necrozele tubulare ischemice sau toxice, boli metabolice, anomalii cogenitale ale rinichiului si tractului urinar cu staza consecutiva intrarenala, nefropatia de reflux, vasculitele sistemice, glomerulonefritele progresive, rinichiul unic congenital/chirurgical, displazia renale unilateral multichistica, etc.) poate fi resposabila de filtrare proteica inadecvata. Proteinuria, legata de mutatii genetice si injuria podocitara, inflamatia, reducerea numarului de nefroni functionali, orice crestere patologica a ratei de filtrare glomerulara, induc modificari hemodinamice la nivelul masei restante de nefroni functionali si determina o stare de stress mecanic cu detasare podocitara si denudarea membranei bazale glomerulare. Aceste mecanisme patologice sunt esentiale pentru formarea aderentelor/ sinechiilor la nivelul capsulei Bowman in glomeruloscleroza. Prezenta proteinuriei este un factor de risc bine cunoscut in progresia bolii renale, in morbiditatea si mortaliatea prin boli cardiovasculare. Scaderea nivelului proteinuriei prin diferite strategii terapeutice diminua aceste riscuri. Prin urmare, este larg acceptat faptul ca reducerea proteinuriei reprezinta o tinta terapeutica in statusul proteinuric cronic. Pe baza studiilor clinice randomizate si controlate s-a concluzionat ca, atat inhibitorii enzimei de conversie ai angiotensinei (ACEI), cat si blocantii receptorilor de angiotensina (ARB), sant, in egala masura, cei mai importanti agenti antiprotenurici si protectori renali. Exista desigur si alte strategii terapeutice si medicatii ce joaca acelasi rol. Dupa cum reiese din numeroasele studii clinice efectuate atat la adult, cat si la copil, tratamentul in aceste cazuri este stratificat pe trei nivele de eficienta: nivelul I (cel mai ridicat), nivelul II (intermediar) si nivelul III (cel mai coborat, in parte experimental). Scopul terapiei anti - proteinurice este scaderea proteinuriei , in mod ideal, la < 300-500 mg/zi.

Concluzii 1.Evaluarea si tratarea proteinuriei cronice la copil inseamna pentru cercetatori si clinicieni un prilej de reevaluare a BRC. 2. Exista evidente solide ca proteinuria reprezinta atat un marker diagnostic cat si un indicator de boala renala progresiva 3. Pentru a preveni complicatiile severe ale BRC din perioada de adolescent-adult tanar este necesara aplicarea precoce a unei strategii terapeutice in toate statusurile proteinurice la copil. 4. Stabilirea unei scheme terapeutice care sa cuprinda medicatii ce modeleaza sistemul renina-angiotensina, controlul TA, indicii de masa corporala, dieta, si in perspectiva tratamente specializate (molecule noi) adresate diferitelor mutatii genetice, are drept scop prezervarea functiei renale pe termen lung in boala renala progresiva sau BRC la copil.

NEW PERSPECTIVES AND STRATEGIES IN PROTEINURIC CHRONIC STATUS IN CHILD

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Proteinuria is not a small problem and represents a diagnosis challenge in pediatric pathology. Persistent proteinuria is a marker of chronic kidney disease (CKD), in adults and children equally. In children, any condition predisposing to glomerulosclerosis (genetic mutations and podocyte injury, severe infections, tubular ischemic or toxic necrosis, metabolic diseases, congenital anomalies of kidney and urinary tract with consecutive intrarenal stasis, reflux nephropathy , systemic

vasculitis, progressive glomerulonephritis, solitary kidney / renal agenesis, unilateral multicystic displastic kidney, etc.), could be responsible of inadequately filtrated proteins. Inflammation, reduced number of functional nephrons, any pathological increased of glomerular filtrated rate, induces hemodynamic adaptable changes in functioning glomeruli and determine mechanical stress with podocyte detachment and glomerular basement membrane (GMB) denudation. These mechanisms are essential for formation of adhesions (synechiae) to the Bowman's capsule in glomerulosclerosis.

The presence of proteinuria is a well-known risk factor for both the progression of renal disease and cardiovascular morbidity and mortality, and decreases in urine protein excretion level were associated with a slower decrease in renal function and decreased in risk of cardiovascular events. It is widely accepted that proteinuria reduction is an appropriate therapeutic goal in chronic proteinuric status.

Based on large randomized clinical trials, ACE inhibitor (ACEI), and angiotensin receptor blocker (ARB) therapy, have emerged as the most important antiproteinuric and renal protective interventions. There are also other clinical strategies and medications that have been shown to be antiproteinuric and therefore renoprotective. There are three levels of complex recommendations according to clinical studies : level 1 (highest), level 2 (intermediate) and level 3 (lowest). The goal of antiproteinuric therapy is to reduce proteinuria ideally to < 300-500 mg/daily.

Conclusion

1. Increasing efforts are being made to prevent and treat CKD in children, before serious complications develop during adolescence and adulthood. 2. It is strong evidence that proteinuria is both a marker for, and a mechanism of kidney disease progression.3. By assessing and treating proteinuria, which is a hallmark of kidney dysfunction, the investigators and clinicians try to find new insights into disease progression and to develop novel treatments for all kidney patients- adults and children 4. The final target of this enterprise is to design studies and strategies that directly engage in manipulation of modifiable factors as renin-angiotensin-system (RAS) interventions, diet, control blood pressure, body mass index, new molecules addressed to the genetic mutations and against podocyte injury, to promote best preservation of renal function in children with progressive renal disease and CKD.

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36. CE SE ASCUNDE IN SPATELE NEFROZEI LIPOIDE LA COPIL?

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Nefroza lipoida sau sindromul nefrotic idiopatic (SNI) este un sindrom de etiologie neprecizata, caracterizat printr-o crestere a permeabilitatii glomerulare urmata de proteinurie masiva, hipoproteinemie, hipogamaglobulinemie si dislipidemie. Expresia clinica a acestor modificari biologice o reprezinta instalarea edemelor masive si a oliguriei.

Etiologia SNI nu este in prezent cunoscuta. Numeroase studii evidenta insa legatura dintre SNI si diatezele atopice. Astfel, aproximativ 50% dintre pacientii cu SNI prezinta semne clinice de atopie cu sau fara cresterea nivelului seric de IgE. Pornind de la această constatare unii autori au încercat o dieta de excludere, care a dus la scăderea proteinuriei, dar nu a redus semnificativ numărul de recidive.

Legatura dintre alimentatie si alterarea barierei intestinale precum si rolul determinant al dietei in reglarea microflorei intestinale si in modularea raspunsului inflamator la nivel intestinal este dovedita in prezent. Prezenta in ser a anticorpilor la alimente de tip IgE, IgG3, IgG4 poate reprezenta un marker de permeabilitate intestinala a crescuta, inflamatie locala si dismicrobism intestinal.

Din experienta noastra, excluderea din dieta pacientilor cu SNI a glutenului, produselor lactate și a alimentelor pentru care exista reacție imună (IgE sau IgG4) a dus la scăderea rapidă a proteinuriei, a permis reducerea duratei totale a corticoterapiei și a împiedicat apariția recidivelor pe o perioadă de un an.

In concluzie, trecerea de la administrarea medicatei antiinflamatoare de tip glucocorticoid la refacerea echilibrului microbiotei intestinale, ameliorarea permeabilitatii intestinale si modularea raspunsului inflamator cu punct de plecare intestinal trebuie sa reprezinte obiectivul principal in managementul SNI la copil.

WHAT LIES BEHIND CHILDHOOD LIPOID NEPHROSIS?

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Lipoid nephrosis or idiopathic nephrotic syndrome (INS) is a syndrome characterized by an increase in glomerular permeability followed by massive proteinuria, hypoproteinemia, hypogammaglobulinemia and dyslipidemia. Clinical expression of these biological changes is the installation of massive edema and oliguria. INS etiology is not currently known. Numerous studies are pointing out the connection between INS and atopic diatheses. Thus, approximately 50% of patients with INS have clinical signs of atopy with or without increasing serum levels of IgE. Based on this finding some authors have tried a diet of exclusion which resulted in decreased proteinuria but not significantly reduced the number of relapses.

There are now numerous clinical studies that highlight the link between diet and impaired intestinal barrier and the decisive role of diet in the regulation of intestinal microflora and in the modulation of intestinal inflammatory response.
The link between diet and altered intestinal permeability and the decisive role of diet in the regulation of intestinal microflora and the modulation of intestinal inflammatory response is currently demonstrated by many authors. The presence in serum of patients with INS of antibodies to food, either IgE, IgG3 or IgG4 may be considered a marker of increased intestinal permeability, local inflammation and intestinal microbiota disturbance.
In our experience, exclusion from the diet of patients with INS of gluten, dairy products and of food for which there is an immune response (sIgE or sIgG4) resulted in a rapid decrease of proteinuria, allowed us to reduce the total length of corticosteroid therapy and prevented relapse of the illness on a period of one year.
In conclusion, changing the treatment protocol from administration of anti-inflammatory medication (glucocorticoid drugs) to restoration of a balanced intestinal microbiota, improvement of intestinal permeability and modulation of the gut inflammatory response must be the primary objective in the management of the child INS.

NEONATOLOGIE / NEONATOLOGY

37. MALFORMATII CEREBRALE, DIAGNOSTIC PRECOCE SI PROGNOSTIC

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Malformatiile cerebrale reprezinta o patologie serioasa in perioada neonatala, cu implicatii majore in dezvoltarea psihomotorie ulterioara a copiilor. Datorita tehnologiilor avansate, diagnosticul este stabilit devreme chiar si in perioada prenatala (ecografie fetala sau chiar mai recent RMN fetal sau RMN in perioada neonatala precoce)- un lucru foarte important pentru introducerea terapiei specifice si de asemenea pentru anticiparea prognosticului pe termen scurt si lung.
Autorii isi propun o trecere in revista a celor mai frecvente malformatii cerebrale: encefalocel, mielomeningocel, malformatia Arnold-Chiari, holoprosencefalia, absenta cavum septum pellucidi, displazia septo-optica, dezordini ale proliferarii si migrarii neuronale, agenezia de corp calos, sindromul Aicardi, porencefalia, defectele liniei mediane, anumite malformatii vasculare.
De asemenea, autorii doresc sa stabileasca o corelatie intre datele clinice, anamnestice si imagistice pentru anticiparea prognosticului pe termen scurt si lung in toate cazurile in care au fost identificate aceste malformatii.
Concluzii: Malformatiile cerebrale reprezinta o parte importanta a patologiei neonatale fie datorita urgentei situatiei cu risc vital fie datorita riscului crescut de sechele cu amenintarea integrarii sociale.
Cuvinte cheie: malformatii cerebrale, diagnostic, prognostic

BRAIN MALFORMATIONS, EARLY DIAGNOSIS AND PROGNOSIS

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The malformations of the brain represent a serious pathology in the neonatal period, with major implications for subsequent psychomotor development of children. Thanks to the advanced tehnology, the diagnosis is established early, even in the prenatal period (fetal ultrasound, or even more recently fetal MRI or early neonatal MRI)-a very important thing for the introduction of the specific therapy and also the anticipation of the short-term and long-term prognosis.
The authors are proposing a review of the most frequent brain malformations: encephalocele, myelomeningocel, Arnold-Chiari malformation, holoprozencephaly, the absence of cavum septum pellucidum, septo-optic dysplasia, proliferation and neuronal migration disorders, corpus callosum agenesis, Aicardi syndrome, porencephaly, median line defects, some vascular malformations
Also, the authors want to establish a corelation between the clinical, anamnestic and imaging data for the anticipation of short-term and long-term prognosis in every group that malformations have been identified.
Conclusion: Brain malformations represent an important part of neonatal pathology, either due to the emergency situation with life-threatening prognosis or because of the high risk of sequelae which threatens social-integration.
Key words: Brain malformation, diagnosis, prognosis

38. PREMATURITATEA EXTREMA- FACTOR DE RISC AL SECHELELOR NEUROLOGICE

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Nasterea prematura , in special prematuritatea extrema este una din cele mai complexe probleme ale medicine moderne. Prin imbunatatirea semnificativa a terapiei intensive neonatale rata de supravietuire a prematurilor cu greutate extrem de mica la nastere creste de asemenea.
Unele din cele mai comune leziuni cerebrale la nou-nascutii prematuri cu varsta de gestatie mai mica de 28 saptamani la nastere sunt hemoragia intraventriculara (IVH)si leucomalacia periventriculara (LPV) . Etiologia hemoragiei intraventriculare este multifactoriala si este atribuita fragilitatii intrinseci ale vaselor matricii germinale si tlburari in fluxul sanguin cerebral.
Leucomalacia periventriculara consta in infarctarea ischemica in regiunea substantei albe cerebrale adiacente ventriculilor laterali. Diagnosticul precoce este stabilit prin ecografia transfontanelara iar aceasta este facuta seriat la toti prematurii internati in clinica noastra. Autorii doresc sa stabileasca incidenta complicatiilor neurologice la prematurii cu greutate extrem de mica la nastere precum si prognosticul pe termen scurt si lung.
Concluzie: In ciuda eforturilor majore in elucidarea patogenezei si prevenirii IVH si LPV din ultimii zeci de ani, prevenirea IVH si LPV ramane o problema nerezolvata. Evaluare neurologica periodica a prematurilor cu greutate extrem de mica la nastere pentru depistarea deficientelor comportamentale si cognitive este necesara pentru a le oferi o speranta de viata cat mai buna.
Cuvinte cheie: prematuritate extrema, complicatii neurologice

EXTREME PREMATURITY – RISK FACTOR OF NEUROLOGICAL SEQUELAE

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Premature birth especially extreme prematurity is one of the most complex problems of modern medicine. By significant improvement of intensive care the survival rate of premature with extremely low birth weight is increasing too. One of the most common cerebral injuries at preterm newborns with gestational age less than 28 weeks at birth are intraventricular haemorrhage (IVH) and periventricular leukomalacia (PVL).The etiology of IVH is multifactorial and is attributed to the intrinsic fragility of the germinal matrix vasculature and the disturbance in the cerebral blood flow. Periventricular leukomalacia consists of an ischemic infarction in the region of the cerebral white matter adjacent to the lateral ventricles. The early diagnosis is established by transfontanelar ultrasonography and this being performed serially to all premature hospitalized in our clinic. The authors want to establish the incidence of neurological complications at extremely low birth newborn and the short-term and long-term prognosis.
Conclusion: Despite major efforts made to elucidate the pathogenesis and prevent IVH and LPV in the last few decades, prevention of IVH and LPV remains an unsolved problem. Periodic neurological evaluation at extreme prematurity for finding behavioural and cognitive impairments is needed for giving them the best expectations in their quality of live
Key words: extreme prematurity, neurological complications

39. ECOGRAFIA DOPPLER ANTE SI POSTNATALA – FACTOR DE PREDICTIE PENTRU RESTRICTIA DE CRESTERE INTRAUTERINA

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Ecografia Doppler la nivelul arterei fetale cerebrale mijlocii (MCA) și arterei ombilicale (UA) reprezinta un factor important de predictie pentru consecintele postnatale in restrictia de crestere intrauterina (IUGR). Ecografia Doppler in al treilea trimestru de sarcină,la sarcinile cu restrictie de crestere intrauterina, prin masurarea indicelui de pulsatilitate(PI) si a indicelui de rezistivitate(IR) la nivelul celor doua artere si postnatal prin ECOTF si Doppler la nivelul arterei cerebrale anterioare sau pericaloase a permis obtinerea unor rezultate catalogate drept negativ majore si negativ minore. Rezultatele negativ majore au fost; decesul perinatal precoce, encefalopatia hipoxic ischemica, hemoragia intraventriculară, leucomalacia periventriculară, enterocolita necrozantă și hemoragia pulmonara. Rezultatele negativ minore au inclus cezariana pentru suferinta fetala, scorul Apgar sub 7 la 5 minute și admiterea in TINN. Concluzii: Studiile Doppler ante si postnale permit monitorizarea fătului compromis și ne pot furniza informatii asupra morbiditatii neonatale in IUGR
Cuvinte cheie: rezultat negativ, Doppler, IUGR

ANTE AND POSTNATAL DOPPLER ULTRASOUND - PREDICTIVE FACTOR FOR INTRAUTERINE GROWTH RESTRICTION

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Fetal middle cerebral artery (MCA) and umbilical artery (UA) Doppler ultrasound is a predictive factor for postnatal consequences of intrauterine growth restriction (IUGR). Doppler ultrasound in the third trimester pregnancies with intrauterine growth restriction, by measuring the pulsatility index (PI) and resistivity index (IR) in the two arteries and postnatally by ECOTF and anterior cerebral artery Doppler or pericaloase allowed obtaining negative results classified as major and minor negative. Major negative results were ;early perinatal death, hypoxic ischemic encephalopathy, intraventricular hemorrhage, periventricular leukomalacia, necrotizing enterocolitis and pulmonary hemorrhage. Minor negative results included cesarean for fetal distress, Apgar score below 7 to 5 minutes and admission TINN
Conclusions: Ante and postnale Doppler studies can monitoring fetal compromise and provide information on neonatal morbidity in IUGR
Key words: negative result , Doppler, IUGR

NEUROLOGIE SI PSIHIATRIE INFANTILA / CHILD NEUROLOGY AND PSYCHIATRY

40. IPOTEZA CELULELOR STEM SI TULBURARILE DE SPECTRU AUTIST

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Tulburarile de spectru autist sunt tulburari de neurodezvoltare care se dezvoltă în primii trei ani de viata caracterizate prin deficit de interacţiune socială, tulburări de comunicare și comportamente restrânse și stereotipe. Prevalența autismului la nivel mondial a crescut foarte mult - in anul 2014, Centrul de Control si Preventie a Bolilor ("Centers for Disease Control and Prevention", SUA) raporteaza ca 1 din 68 de copii cu varsta de 8 ani sufera de o Tulburare de Spectru Autist.
Etiologia Tulburarilor de spectru autist nu este inca cunoscuta, implicand cel mai probabil o interactiune a factorilor genetici, imunologici, și de mediu. Studiile au aratat ca in TSA este afectata procesarea informatiei la nivel cerebral prin alterarea organizarii si conectivitatii sinaptice.
Pornind de la aceste observatii, cercetatorii au propus o metoda inovativa de tratament: terapia cu celule stem. Studiile anterioare pe animale au aratat capacitatea acestor celule de a stimula refacerea tecii de mielina si a conectivitatii celulelor din maduva spinarii. Desi deocamdata ramane neclar modul in care celulele stem recoltate din cordonul ombilical pot influenta conectivitatea neuronala cerebrala din autism , cercetatorii anticipeaza ca anumite subtipuri de autism pot beneficia de pe urma acestei terapii.
Cuvinte cheie: Tulburare de spectru autist, ipoteza celulelor stem

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Autism spectrum disorders are neurodevelopmental disorders that develops in the first three years of life characterized by deficient social interaction, impaired communication, restricted and stereotyped behaviors. Global prevalence of autism has greatly increased - in 2014, the Center for Disease Control and Prevention ("Centers for Disease Control and Prevention ", USA) reported that 1 of 68 children, 8 years old is suffering from an Autistic Spectrum Disorder.
The etiology of autism spectrum disorder is yet unknown, most likely involving an interaction of genetic, immunologic, and environmental factors. Studies have shown that in TSA the information processing in brain is affected by the alteration of synaptic organization and connectivity.
Based on these observations, researchers proposed an innovative method of treatment: stem cell therapy. Previous studies on animals have shown the ability of these cells to stimulate the recovery of myelin sheath and connectivity cells in spinal cord. Although it remains unclear how stem cells collected from the umbilical cord may influence brain neuronal connectivity in autism, researchers anticipate that certain subtypes of autism can benefit from this therapy.
Keywords: Autism Spectrum Disorder, stem cell hypothesis.

41. ABORDAREA EDEMULUI CEREBRAL ACUT IN PRACTICA PEDIATRICA

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Autorii prezinta date referitoare la definitia, etiologia si fiziopatologia edemului cerebral acut. Se trec in revista multiplele cauze care pot determina cresterea presiunii intracraniene si anume cresterea continutului in apa al tesutului cerebral (acesta fiind diferit pentru substrata alba si substanta cenusie), infectiile virale cu tropism neurologic, traumatisme cranio-cerebrale, in cadrul hipertensiunii arteriale, in jurul tumorilor cerebrale, a zonelor de ischemie, infarctizare sau trombozelor. Hipertermia, ca si expunerea prelungita la soare pot determina, de asemenea, cresterea presiunii intracraniene. Referitor la clasificare, se iau in discutie diversele tipuri fiziopatologice de edem cerebral acut (vasogenic – lezarea barierei hematoencefalice sau citotoxic – modificarea metabolismului celulei nervoase). In continuare, autorii analizeaza simptomatologia neurologica si investigatiile paraclinice cu utilitate diagnostica pentru a elucida cauza cresterii presiunii intracraniene. In finalul lucrarii se fac referiri la tratamentul fiecarei forme de edem cerebral acut, in fuctie de cauza si evolutia specifica fiecarei etiologii.

NUTRIŢIE ŞI BOLI METABOLICE / NUTRITION AND METABOILIC DISEASE

42. THE ROLE OF GLUTEN IN THE PATHOGENESIS OF TYPE 1 DIABETES MELLITUS AND CROHN DISEASE

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The role of gluten is well known in the pathogenesis of coeliac disase,as its trigger factor. Gluten is characterized by its high content of prolin and glutamin, and after their digestion even by the normal small intestinal mucosa long polypeptides remain, which are able to increase the permeability of even normal intestinal mucosa through their ability to release zonulin. It can be stated, that gluten could have a dose-dependent detrimental effect on the gut barrier, primarily in the small intestine even without coeliac disease. This effect of gluten raises the possibility its pathogenetic role of other diseases in addition to coeliac disease, first of all in type 1 diabetes mellitus (T1DM) and Crohn disease (CD).

Our earlier observation that in the jejunal mucosa of children with T1DM signs of immune activation are observed could have been the consequence of increased intestinal permeability. In this study the patients had significantly more α4/β7 integrin positive cells in the lamina propria compared to controls. This observation is highly relevant, because T-cells from humans diabetic pancreas with T1DM express the gut-associated homing receptor α4/β7 integrin. The pathogenetic role of gluten in induction of T1DM is indicated also by the frequent association of T1DM and coeliac disease and it was observed that in the majority of cases the onset of T1DM is earlier than that of coeliac disease, when the patient is not on a gluten free diet.

According to several studies it is probable that beside characteristic genetic traits mostly the diet influences the development of CD. The prevalance of CD is on the rise in developed countries exposed to Western diet. This is also indicated by the very efficient therapeutical results of exclusive enteral nutrition (EEN) in CD. Pediatric studies confirmed that 70-90% of children fed an exclusive liquid formula and no exposure to other food, reach a complete remission. The effect of formula is not depend on its composition or the degree of hydrolysis of its protein (elemental or polymer formula). Therefore, the EEN may have its effect principally that it does not contain those compounds which are frequently occur in the normal diet, it is also gluten free. Increased gut permeability caused by gluten may be the primary factor triggering CD.

On the base of the above mentioned fact it is probable that gluten may play a decisive role in the pathogenesis of type 1 diabetes mellitus (T1DM) and of Crohn's disease (CD).

43. NUTRITIA IN OBEZITATEA COPILULUI

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Obezitatea copilului se defineste ca o afectiune cronica complexa, multi-factoriala. Nu exista un consens in ceea ce priveste tratamentul copilului obez, de aceea, preventia reprezinta abordarea ideala. Daca preventia nu da rezultate, este necesara interventia nutritionala. Rezultatele din ultimii ani arata ca nutritia in perioada postnatala precoce si, ulterior in

copilarie, poate reprezenta principalul mijloc de preventie in obezitate. In perioada postnatala precoce, exista factori (alaptarea si diversificarea alimentatiei) importanti pentru riscul dezvoltarii excesului ponderal, ulterior in copilarie. S-a dovedit ca alaptarea reduce riscul de obezitate la copil. Aportul crescut de proteine in cursul diversificarii este asociat cu un risc mai crescut de obezitate, in timp ce aportul crescut de lipide in aceasta perioada, nu pare sa reprezinte un factor de risc pentru obezitate, mai tarziu. Dimpotriva, la copilul mai mare si adult, aportul crescut de lipide este asociat cu un risc mai inalt de obezitate. Castigul ponderal rapid in perioada postnatala precoce (0 - 6 luni), reprezinta un indicator sensibil pentru riscul ulterior de obezitate. Acolo unde preventia nu a fost eficienta, interventia nutritionala este necesara pentru managementul obezitatii. Exista o serie de recomandari generale standard, dar si modele dietetice speciale (diete cu: continut redus/mediu de glucide, cu indice glicemic redus, cu valoare energetica redusa) care pot fi utilizate cu succes in managementul copilului obez. Concluzii. Nutritia reprezinta, de la cele mai fragede varste, elementul esential in preventia obezitatii la copil; interventia nutritionala in obezitatea copilului trebuie adaptata varstei, tinand cont ca, copilul este un organism in crestere.

NUTRITION IN CHILD OBESITY

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Child obesity is a complex, multi-factorial, chronic medical pathology. There is no consensus in treating child obesity, so prevention remains still an ideal approach. If prevention did not succeed, than nutritional intervention is needed. In the last years, there is evidence that nutrition in early infancy and childhood might be used as a preventive tool in the management of obesity. Some factors in early life are important for the risk of overweight and obesity later in childhood. Among the postnatal factors, breastfeeding and complementary feeding are of interest. Breastfeeding has been shown to reduce the risk of later obesity. A high protein intake during complementary feeding is associated with a higher risk of obesity while, a high fat intake during this period does not seem to be a risk factor for later obesity. On contrary, in older children and adults, a high fat intake is associated with higher risk of obesity. Rapid weight gain early postnatal (0 - 6 months), seem to be a good indicator for the risk of obesity later in life. If prevention does not succeed than dietary intervention would be the second tool in management. Some standard general recommendations and also some special dietary models (low/medium carbohydrate diet, energy restricted diet, low glycaemic index diet) can be used to treat overweight/obese children. Conclusion. Nutrition even since early childhood is essential to prevent later obesity; nutritional intervention should be adapted to age and need to keep in mind that the child is continuously growing.

44. OLIGOELEMENTELE - ROLUL LOR ÎN ÎN NUTRIȚIE

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Oligoelementele (fier, zinc, cupru cobalt, mangan iodul, seleniu, molibden, crom și fluor), au roluri bine definite la copii, atât pentru procesul de creştere, cât şi pentru menţinerea stării de sănatate. Acestea se mai numesc și microminerale esențiale. Cantitatea necesară zilnică e de ordinul a câteva miligrame pe zi. Pe lângă acestea, siliciul, staniul, nichelul,vanadiul, litiul, arsenicul, borul, germaniul, cadmiul, plumbul, rubidiul și argintul, aflate în organism în concentraţii mai mici decât precedentele, au implicaţii biologice importante. Aportul de microelemente este asigurat prin ingestia de alimente de origine animală şi vegetală, dar şi prin alimentele fortifiate sau suplimentele alimentare. Pediatrul trebuie să cunoască recomandările privind aportul normal de oligoelemente în funcţie de diferitele categorii de vârstă și să poată estima corect necesitatea suplimentării în scop profilactic sau terapeutic. La sugar, înțarcarea e un moment critic în ceea ce priveşte aportul de oligoelemente. De asemenea, în adolescenţă necesarul zilnic din anumite oligoelemente, alături de macrolemente şi vitamine este mai mare decât în orice altă perioadă a vieţii. Dorinţa de suplimentare cu unii micronutrienţi (vitamine şi/sau oligoelemente) poate avea efecte negative. Cunoaşterea efectelor adverse ale excesului de macroelemente, oligoelemente şi vitamine, precum şi interacţiunile dintre membrii acestor clase de nutrienţi e la fel de importantă ca recunoaşterea şi tratarea carenţelor. Cuvinte cheie: oligoelemente, nutriţie, copil

THE TRACE ELEMENTS AND THEIR ROLE IN NUTRITION

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The trace elements (iron, zinc, copper, cobalt, manganese, iodine, selenium, molybdenum, chromium and fluoride), children have defined roles for both growth process and for maintaining health. They are also called essential trace minerals. The daily intake requirements are about few milligrams per day. In addition, silicon, tin, nickel, vanadium, lithium, arsenic, boron, germanium, cadmium, lead, rubidium, and silver are found in lower concentrations than the previous ones, but have important biological implications. The intake of micronutrients is provided by food of animal and vegetable origin, but also through fortified foods or supplements. The pediatrician should know the normal intake recommendations of trace elements in the different age categories and to estimate correctly the need to supplementation for prophylactic or therapeutic purposes. In infants, weaning is a critical time in terms of micro-nutrient intake. Also in adolescence, the daily requirement of certain

trace elements and vitamins along with macro elements is higher than in any other period of life. The unnecessary supplementation with some micronutrients (vitamins and / or trace elements) can have negative effects. Understanding the adverse effects of excess macro, trace elements and vitamins, and the interactions between members of these classes of nutrients is as important as recognizing and treating deficiencies. Keywords: trace elements, nutrition, child

45. NUTRITIA SI CREIERUL

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Cresterea, dezvoltarea si maturarea cerebrala optima presupune o nutritie adecvata. Dezvoltarea cerebrala prenatala precoce si din primii ani de viata depinde de asigurarea corespunzatoare a unor factori nutritionali cheie. Cu toate ca, toti nutrientii sunt importanti pentru o dezvoltare cerebrala normala, doar unii dintre acestia (proteine, acizi grasi polinesaturati cu lant lung, fier, zinc, cupru, iod, acid folic, colina si vitaminele A, B6, B12) prezinta efecte semnificative in primii ani de viata, caracterizati prin dezvoltarea rapida a anumitor regiuni cerebrale care implica nevoi crescute de nutrienti.
Agresiunile nutritionale precoce exercita un efect seminificativ asupra proliferarii celulare, disctinct de cele tardive care afecteaza predominant diferentierea, inclusiv sinaptogeneza si arborizarea dendritica.
Nutritia este un factor de mediu care interfera cu nivelele tisulare ale neurotransmitatorilor si determina modificari neuroanatomice, neurochimice si neurometabolice.
Consecintele functionale ale acestor modificari variaza in functie de tipul deficitelor nutritionale specifice si de momentul de actiune al acestora in raport cu procesul de crestere si dezvoltare cerebrala.
Cresterea cerebrala este foarte rapida in primii 2 ani de viata, indeosebi la nivel cortical si atinge nivelul greutatii adultului la varsta de 6-14 ani. Mielinizarea cerebrala este mai accentuata in ultimele doua trimestre de sarcina si in primii 2 ani de viata, continuand pana la varsta pubertatii.
Nutritia exercita efecte directe asupra expresiei genelor la nivel cerebral prin alterarea acetilarii histonelor si a expresiei factorilor de crestere neuronala.
Astfel, nutritia detine un rol critic in interactiunea factorilor biologici si nutritionali care mediaza cresterea si dezvoltarea cerebrala.
Cuvinte cheie: copii, nutritie, crestere si dezvoltare cerebrala

NUTRITION AND BRAIN DEVELOPMENT

Dr. Victoria Hurduc

The brain requires adequate nutrition for optimum growth, development and maturation. Optimal overall brain development in the prenatal period and early years of life depends on providing sufficient quantities of key nutrients. Wile all nutrients are important for brain development, certain nutrients (protein, long chain polyunsaturated fatty acids, iron, copper, zinc, iodine, folate, choline and vitamin A, B6 and B12) have particularly large effects early in life, when specific brain regions are developing most rapidly and have highest nutrient requirements.
For any given region, early nutritional insults have a greater effect on cell proliferation, thereby affecting cell number. Later nutritional insults affect differentiation, including synaptogenesis and dendritic arborization. Nutrition is an environmental factor that can interfere with the tissue levels of neurotransmitters, resulting in neuroanatomical, neurochemical or neurometabolic changes.
The functional consequences of these alterations vary, depending on the specific nutritional deficiency and the timing of the deficiency relative to the growth and developing neurological processes.
Brain weight is very rapid during the first 2 years of life, particularly in the cortex and achieves adult weight between 6 and 14 years of age. The myelination of the brain is concentrated from mid-gestation through the second year of life, but continues through puberty.
Nutrition can have direct effects on gene expression in brain by altering histone acetylation and the effects of hypoglycemic diets on the genetic expression of neuronal growth factors.
Thus, nutrition plays a critical role at the cross-roads of the biological and nurturing factors that mediate brain growth and development.
Keywords: children, nutrition, cognition, neurogenesis

46. ALIMENTATIA COMPLEMENTARA SI MICROBIOMUL

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Microbiomul are roluri importante in organismul uman: protectia impotriva agentilor patogeni, prelucrarea de nutrienti, stimularea angiogenezei si reglarea mecanismelor de stocare a grasimilor. Colonizarea tubului digestiv are loc in paralel cu maturizarea sistemului imun si intervine in mentinerea functionarii normale a acestuia. Formarea microbiomul intestinal uman incepe inca inainte de nastere si este un proces dinamic.

Au fost identificate două aspecte importante in privinta modului in care alimentatia influenteaza dezvoltarea microbiomului intestinal la copii: alăptarea care, indiferent de durata, determina un echilibru bacterian intestinal unic si diferit de cel observat la indivizii care consuma alimente solide; si alimentatia complementara cu rol semnificativ mai mare in modelarea diferitelor prototipuri de microbiom intestinal pe termen lung, comparativ cu alti factori de mediu (De Filippo et al., 2010). Perioada de sugar este cea mai labila din punct de vedere al modificarii microbiomului comparativ cu alte etape de dezvoltare.

In timp ce bolile alergice reprezinta cele mai comune boli cronice ale copilariei, obezitatea este cea mai raspandita problema de nutritie in tarile occidentale. Pornind de la aceasta constatare, se studiaza manipularea microbiomului in aceasta perioada importanta din viata in scopul minimalizarii efectelor acestor afectiuni.

Manipularea microbiomului uman se poate realiza cu suplimente alimentare cu microorganisme vii ("bacterii benefice" - probiotice si simbiotice), cu prebiotice si o dieta bogata in oligozaharide si prin antibiotice (medicatie care distruge/suprima microorganismele vii din intestin). Studiile in curs de desfasurare ar putea conduce la strategii de manipulare a microbiomului intestinal in vederea imbunatatirii starii de sanatate a populatiei pediatrice.

COMPLEMENTARY FEEDING AND THE MICROBIOME

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This microbial ecosystem serves numerous important functions for its human host, including protection against pathogens, nutrient processing, stimulation of angiogenesis, and regulation of host fat storage. Microbial colonization runs in parallel with immune system maturation and plays a role in intestinal physiology and regulation. Increasing evidence on early microbial contact suggest that human intestinal microbiota is seeded before birth.

Two important points related to dietary drivers of the gut microbiome development in children were identified. First, breast-feeding, regardless of duration supports a specific bacterial state that is unique and markedly different from that observed in individuals consuming solid foods. Second, once solid foods are introduced, its role in shaping long-term gut microbiome profiles is so strong that individual's cluster based on diet type over other environmental and physiological factors (De Filippo et al., 2010). During infancy the intestinal microbiota is less stable and more variable in its composition than in older children and adults.

Allergic diseases comprise the most common chronic disease in childhood while obesity is the most prevalent nutritional problem in Western countries. As diet plays a major role in the development of microbiota during this period, there is an opportunity for its manipulation.

Manipulation of the human microbiome may include microbial supplements (probiotics or synbiotics), foods or substrates (diet or prebiotics), and microbial suppression or elimination,Future studies may lead to improved health benefits for pediatric patients through the manipulation of the intestinal microbiota (antibiotics) strategies.

PNEUMOLOGIE / PNEUMOLOGY

47. PARTICULARITATILE ASTMULUI SEVER LA COPIL

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Astmul este cea mai frecventa boala cronica a copilului si o importanta problema de sanatate in intreaga lume.
Severitatea astmului este cea mai importanta trasatura a bolii fiind responsabila de consecintele pe termen scurt si lung ale acestuia.
Initial severitatea astmului era stabilita pe baza simptomelor, necesarului de medicatie de tip reliever si a testelor functionale respiratorii.
Cele mai recente ghiduri privind managementul astmului recomanda stabilirea severitatii pe baza step-ului de tratament care asigura controlul bolii.
Inainte de a decide ca un pacient are astm sever este important sa se faca deosebirea intre astmul sever si astmul dificil de tratat datorita unei tehnici inhalatorii incorecte, lipsei de complianta sau comorbiditatilor incomplet tratate.
Astmul sever la copil are o serie de particularitati fata de astmul sever al adultului.

La copil astmul sever este strans legat de atopie spre deosebire de astmul adultului.
Adolescentii au cea mai mare prevalenta a astmului sever si cel mai mare risc de deces prin astm datorita compliantei scazute la tratament si a comportamentelor la risc (fumatul).
La copiii cu astm sever rezultatele testelor functionale respiratorii sunt dependente de varsta si pot fi in limite normale in ciuda prezentei simptomelor in momentul evaluarii.
Astmul sever la copil are o etiologie multifactoriala. Exista mai multe particularitati genetice, epigenetice si moleculare asociate astmului sever la copil.
Principalele directii ale managementului astmului sever la copil sunt optimizarea terapiei si tratamentul comorbiditatilor.
Perspectievele in ceea ce priveste tratamentul astmului sever la copil includ individualizarea acestuia si utilizarea terapiilor biologice.
Cuvinte cheie: boala respiratorie cronica, astm sever, copil

PECULARITIES OF SEVERE ASTHMA IN CHILDHOOD

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Asthma is the most common chronic disease in children and an important health problem worldwide. Asthma severity is the most important feature of asthma being related to its short and long term outcomes.
At first asthma severity was established according to the level of symptoms, the need for rescue medication and lung function tests.
Most recent guidelines for asthma management recommend that asthma severity should be determined according to the step of therapy needed to achieve asthma control.
Before deciding whether a patient has severe asthma or not, it is important to distinguish between severe asthma and difficult to treat asthma related to incorrect inhaler technique, improper adherence or incomplete controlled comorbidities.
Severe asthma in children has a series of distinctive features towards severe asthma in adults.
In children severe asthma is closely linked with atopy unlike severe asthma in adults.
Adolescents are at a highest prevalence of severe asthma and at the highest death risk through asthma do to poor treatment adherence and do to at risk behaviors (smoking).
In children with severe asthma lung function measurements are age dependent and might be between normal ranges despite the presence of symptoms at the time of the assessment.
Severe asthma in children has a multifactorial etiology. There are many molecular, genetic and epigenetic patterns related to severe asthma.
The main drivers of severe asthma management in children are therapy optimization and comorbidities treatment.
The perspectives in severe asthma management in children include individualized treatment and biological therapies.
Key words: chronic respiratory disease, severe asthma, child

48. COMPLICATED PNEUMONIA IN CHILDREN

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Community-acquired pneumonia (CAP) is an important cause of morbidity and mortality in children worldwide. Pneumonia with pleural fluid (PF) and empyema are known complications of CAP, especially when a bacterial etiology is present, with effusions occurring in at least 40% of bacterial pneumonia episodes.
Differentiating PF from all other pneumonia (Pn) episodes mainly relies on radiographic diagnosis, it is difficult to distinguish between the two, based only on clinical symptoms at the time of admission. In children with complicated pneumonia, a more severe clinical presentation with longer disease and hospitalization duration was reported compared to children with non-complicated pneumonia. PF illness was associated with higher rates of prolonged fever, tachypnea and abdominal pain; elevated inflammatory markers, such as C-reactive protein (CRP) and thrombocytosis; as well as higher rates of hypoalbuminemia and hyponatremia when compared to children with non-complicated pneumonia. Furthermore, slow and unsatisfying response to antibiotic treatment has been reported in complicated pneumonia patients.
The most common pathogens of complicated pneumonia in children are *Streptococccus pneumoniae*, *Staphylococcus aureus* and *Haemophilus influenzae*, while the major viral pathogens are influenza-virus, parainfluenza and respiratory syncytial virus (RSV).
Treatment can be conservative based on antibiotic adequate treatment and supportive treatment in cases when small amount of pleural fluid exist. However, when there is a large amount of pleural fluid and in cases of respiratory de-compensation, insertion of a chest drain is an important treatment and can be inserted in most cases under ultrasound and in some cases under CT scan. The new pneumococcal conjugated 13-valent vaccine reduced significantly the rates of complicated pneumonia in several countries.

49. UPDATE IN PAEDIATRIC RESPIRATORY FAILURE

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Abstract

Respiratory failure is a paediatric emergency condition. It is defined as an inability of the respiratory system to provide sufficient oxygen for metabolic needs (type 1 or hypoxemic respiratory failure) or to excrete the CO_2 produced by the body (type 2 or hypercapnic or ventilatory respiratory failure). Epidemiological data shows that respiratory diseases account for around 10% of all visits to paediatric emergency departments, though majority of them are mild and self-limiting. But 20% of its represents hospital admissions. Infants and young children develop respiratory failure faster compared to older children and adults. Many different disease processes lead to respiratory failure. Respiratory failure is the end of acute or chronic progression of respiratory illness and distress Several predisposing factors increase risk for developing respiratory failure in infants and children. During hypoxia a paradoxical inhibition of respiratory drive with apnoea or hypoventilation instead of hyperventilation ensues. Therefore recognizing the symptoms and signs of impending respiratory failure is important.

An ABCD approach to infants and children with respiratory failure is an established urgent approach in paediatric emergency medicine. Many times respiratory failure occurs in the out-hospital settings where trained medical or paramedical teams in paediatric emergencies are of utmost importance. In both settings, in-hospital and out-hospital medical teams should be available to manage difficult airway management. An algorithm how to approach to difficult airway management should be a part of every emergency medical team. A decision to start non-invasive ventilation depends on aetiology of respiratory diseases and clinical condition at impending respiratory failure as well as possibilities and capabilities of transporting and paediatric emergency team to start with it. Mechanical ventilation after successful endotracheal intubation or introducing newly supportive artificial airways-laryngeal masks of different types can be started only after proper sedation and analgesia which prevent extubation or removal of laryngeal masks. Different modes of ventilation are available which together with advancing in computer technology enables better ventilator-patient coordinate and synchronous breathing, among theme are neurally adjusted ventilation, NAVA and proprotional assist ventilation, PAV. High frequency oscillatory ventilation is also promising in decreasing lung trauma. Better knowledge and understanding of pathophysiology of the diseases of respiratory system as well as it interactions and interplay with other organs together with better cardio-respiratory monitoring help health care workers to better adapation and correction of all necessary key cardiorespiratory parameters according to the on-going needs of the patients. Preventive and pre-emptive measures are helpful in some diseases preventing them to progress to respiratory failure. Antibiotics, monoclonal antibodies to prevent virus replication, novel biological drugs modulating immune response, earlier correction of congenital heart defects, etc. are indispensable armamentarium of modern neonatal and paediatricintensive care. Respiratory gases, nitrogen oxide, NO, heliox and the most advanced technologies as extracorporeal membrane oxygenation systems help in the severe hypoxic respiratory failure as well as hyperbaric oxygen chamber in some specific conditions. In rare cases lung or heart-lung transplantation may save the life of child. Rarely chronic home mechanical ventilation is needed if chronic after acute respiratory failure ensues. Modern home mechanical ventilators are capable working on batteries for many hours which allows the patients are more movable and less dependent to stay near electrical power all the time.

In conclusion we may confirm that progress in understanding the patophysiology of respiratory failure together with better medicinal products and equipment and rapid response emergency teams in- and-out the hospitals may help in better outcomes of the patients with acute respiratory failure, though still many questions remained opened and to be resolved.

50. PNEUMOPATIILE INTERSTIȚIALE CRONICE LA COPIL

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În ultima decadă s-au înregistrat progrese importante în domeniul pneumopatiilor interstițiale cronice la copil determinate de descoperirile în domeniul geneticii, ameliorarea tehnicilor de imagistică pulmonară și eforturilor de a defini mai bine fenotipurile clinice. Aceste progrese au dus la elaborarea unei noi clasificări a acestor entități, clasificarea utilizată la adulți nefiind potrivită spectrului pediatric. Luate global aceste boli sunt rare la copil (1,32 - 3,6 cazuri/1 milion populație), de aici derivă și lipsa de informare a pediatrilor cu privire la acest capitol. Tabloul clinic și paraclinic constă în tuse, polipnee, raluri crepitante, falimentul creșterii și hipoxemie. Wheezing-ul este mai rar întâlnit. Diagnosticul acestor boli presupune în primul rând excluderea unor entități de suferință respiratorie cronică mai frecvente: fibroză chistică, imunodeficiențe, cardiopatii congenitale, displazie bronhopulmonară, infecții respiratorii, diskinezie ciliară primară sau aspirația recurentă. Principalele examinări paraclinice utilizate în diagnosticul pneumopatiilor interstițiale cronice sunt: 1) imagistica pulmonară (radiografie toracică, tomografie computerizată, tomografie computerizată de înaltă rezoluție); 2) probe funcționale

respiratorii; 3) bronhoscopie cu lavaj bronhoalveolar; 4) teste genetice; și 5) biopsie pulmonară. Prognosticul variază în funcție de boală. Chiar luate global prognosticul acestor pneumopatii interstițiale cronice este variabil: de la 64% supraviețuire la 5 ani la 94% supraviețuire. Arsenalul terapeutic este limitat și constă în: 1) imunospupresoare (corticosteroizi, hidroxiclorochină); 2) terapie de susținere (nutriție adecvată, oxigenoterapie, ventilație non-invazivă sau invazivă), 3) evitarea factorilor agravanți (evitarea fumatului pasiv, vaccinări, imunoglobuline anti-VRS) și 4) transplant pulmonar.
Cuvinte cheie: pneumopatie interstițială cronică, copil

CHILD INTERSTITIAL LUNG DISEASES

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In the last decade there have been major advances in the field of child interstitial lung diseases due to discoveries in genetics, improved pulmonary imaging techniques and efforts to better define clinical phenotypes. These advances have led to a new classification of these entities, the classification used in adults is not suitable for pediatric spectrum. Overall these diseases are rare in children (1.32 to 3.6 cases / 1 million population), hence derives pediatricians' lack of information on this chapter. Clinical and laboratory manifestations consists of cough, tachypnea, crackles, failure to thrive and hypoxemia. Wheezing is less common. The diagnosis of these diseases primarily involves the exclusion of common chronic respiratory disease: cystic fibrosis, immunodeficiencies, congenital heart disease, bronchopulmonary dysplasia, respiratory infections, primary ciliary dyskinesia or recurrent aspiration. The main laboratory examinations used in the diagnosis of interstitial lung diseases are: 1) pulmonary imaging (chest X-ray, computed tomography, high-resolution computed tomography); 2) pulmonary function tests; 3) bronchoscopy with bronchoalveolar lavage; 4) genetic tests; and 5) lung biopsy. The prognosis varies depending on the disease. Even taken overall prognosis of these interstitial lung diseases is variable: from 64% survival at 5 years to 94% survival. Therapeutic arsenal is limited and consists of: 1) immunosuppressive drugs (corticosteroids, hydroxychloroquine); 2) supportive therapy (proper nutrition, oxygenation, non-invasive or invasive ventilation), 3) avoiding aggravating factors (avoiding passive smoking, vaccinations, anti-RSV immunoglobulin) and 4) lung transplant.
Key words: interstitial lung disease, child

REUMATOLOGIE / REUMATHOLOGY

51. FEBRA PERIODICA LA COPIL

Mariana Stefan
Regina Maria

Febra este cauza principala care aduce copilul la medic, iar etiologia acesteia tine de toate subspecialitatile pediatrice. Diagnosticul etiologic trebuie sa excluda cauzele infectioase, oncologice, dermatologice, endocrinologice... Daca aparitia febrei este repetitiva, este de luat in discutie originea reumatologica. Afectiunile reumatologice in care apare „ febra recurenta", mai corect etichetata decat „periodica" sunt descrise in anii 80 si apartin de categoria afectiunilor autoinflamatorii. Bolile autoinflamatorii sunt reprezentate de un numar de afectiuni ce apar secundar mutatiilor la nivelul unei gene, care codifica proteine cu rol esential in reglarea raspunsului inflamator. Dintre acestea se vor trece in revista cele incadrate in categoria „ febrelor periodice" si cele asociate cu febra.
Scopul prezentarii este de a indemna pediatrii sa ia in calcul la stabilirea etiologiei febrei si cauzele reumatismale.

PERIODIC FEVER IN CHILDREN

Mariana Stefan
Regina Maria

Fever is the main cause that bring the child to doctor. Etiology belongs to almost all pediatric subspecialities. The etiologic diagnosis should exclude infectious, oncologys, dermatologys, endocrinologys causes. If fever is recurrent, we can take into considerations the rheumatological origins.
Conditions known as „ periodic fever" are rather „recurrent fever" and have been described during 80s and belong to the autoinflamatory diseases. The autoinflamatory diseases are represented by a number of conditions that occur secondary to mutations in a gene that encodes proteins with an essential role in regulating the inflamatory response. Among all the autoinflamatory conditions, we will discusse those belonging to the category of „periodic fever" or associated with it.
Goal of the paper is to urge pediatricians to consider the rheumaic fever in determining fever etiology.

52. URGENȚE ÎN BOLILE REUMATISMALE LA COPIL

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Uneori evoluția cronică a bolilor reumatismale poate fi întreruptă de complicații cu risc vital imediat precum sindromul de activare macrofagic (SAM) și sindromul antifosfolipidic catastrofal (SAF). Diagnosticul cat mai precoce completat de un tratament adecvat este esential în astfel de situații. Opțiunile terapeutice în ambele situații sunt în relație cu patogenia imună și vizează anihilarea rapidă și agresivă a perturbarilor legate de hiperproducția citokinică (TNF a, IL1, IL6,IL2,IFNg, GM-CSF) şi activitatea LT- NK ,LT-CD8 pentru SAM și anomaliile de coagulare de tip trombotic în cazul SAF declansate pe de o parte de dereglarea apoptozei celulare în cadrul inflamației imune cu formarea secundara de Ac anticardiolipina, Ac anti b2-glycoproteina1, anti- factori de coagulare (proteina C, S, protrombină, anexine) iar pe de alta parte de activarea concomitentă plachetară , a endoteliului vascular și a sistemului complement. Nu exista încă un protocol terapeutic utilizabil numai la copil cel folosit în prezent fiind preluat de la adult. Astfel pentru SAM tratamentul cuprinde doze mari de corticosteroizi, inhibitori de calcineurina ,inhibitori de TNFa si IL1. SAF beneficiază de anticoagulante (G moleculara mica) asociate cu anti agregante plachetare , Ac anti-CD20 , Ac anti C5, Ig iv, plasmafereză, corticoterapie, ciclofosfamidă. Eficiența terapeutica va fi cuantificată prin markeri biologici precum raportul feritina serica/feritină glicozilată, CRP, VSH , ameliorarea disfuncției organice și normalizarea coagularii. **În concluzie,** SAM și SAF catastrofal reprezintă urgențe în cadrul bolilor reumatismale ce afecteaza prognosticul vital al bolnavului și care necesita diagnostic rapid , tratament și îngrijire în echipă multidisciplinară.

EMMERGENCY IN RHEUMATOLOGIC DISEASE IN CHILDREN

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Sometimes the chronic rheumatic diseases progress can be interrupted immediately by life-threatening complications such as macrophage activation syndrome (MAS) and catastrophic antiphospholipid syndrome (CAPS). The early diagnosis completed with appropriate treatment is essential in such situations. Therapeutic options in both cases are related to the immune pathogenesis and targeting the rapid and aggressive annihilation of the disturbances related hyper-cytokine production (TNF-a, IL1, IL6, IL2, IFNg, GM-CSF) and LT- NK , LT-CD8 activity for MAS and the coagulation abnormalities expressed by thrombotic events in the case of CAPS triggered on the one hand by the upregulation of cellular apoptosis secondary of immune inflammation and production of Ab anticardiolipin , anti b2-glycoprotein1, anti-coagulation factors (protein C, S, prothrombin, annexin) and on the other hand by the co-activation of platelets, vascular endothelial and complement system. There are no therapeutic protocol still has used only at the child the actually being the taken from adult.Thus, for MAS treatment includes high doses of corticosteroids, calcineurin-, TNFa- and IL-1 inhibitors. APS benefit from the anticoagulants (low molecular G) associated with anti-platelet aggregates, anti-CD20, anti C5, iv Ig, plasmapheresis, corticosteroids, cyclophosphamide. Therapeutic efficiency will be measured by the biological markers such as serum ferritin ratio / glycosylated ferritin, CRP, ESR, improved the organ dysfunction and the coagulation normalization. In conclusion, MAS and CAPS are the catastrophic emergencies in the rheumatic child diseases affecting the vital prognosis of the patient and which require rapid diagnosis, treatment and care in a multidisciplinary team.

53. ASPECTE DIAGNOSTICE SI TERAPEUTICE ÎN VASCULITELE SISTEMICE LA COPIL

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Vasculitele sistemice pediatrice reprezinta un grup de afectiuni cu incidenta redusa, avand manifestari clinice heterogene si ridicand probleme dificile de diagnostic si tratament. Elementul comun il reprezinta inflamatia peretelui vascular, in unele forme asociind si granuloame necrotizante extravasculare. Manifestarile clinice sunt reprezentate atat de simptome generale, dar si de simptome specifice organelor afectate.

Autorii discuta modificarile survenite in nomenclatura, clasificarea si criteriile de diagnostic ale consensurilor internationale, incepand cu Chapel-Hill (1994 si 2012) si continuand cu criteriile propuse de EULAR/PRINTO/PRES in 2010, acestea din urma fiind rezultatul unui grup international recunoscut ca lider european in reumatologia pediatrica. Grupurile reunite EULAR/PRINTO/PRES au validat criterii de diagnostic pentru principalele patru vasculite pediatrice: purpura Henoch-Schonlein, panarterita nodoasa a copilului, granulomatoza Wegener, si arterita Takayasu. Este prezentat succint si scorul de evaluare a activitatii vasculitei copilului - PVAS (Paediatric Vasculitis Activity Score), precum si optiunile terapeutice disponibile in acest moment.

Autorii subliniaza ca, in pofida progreselor diagnostice si terapeutice inregistrate in ultimele doua decenii, vasculitele sistemice ale copilului au in continuare un grad crescut de subdiagnosticare de catre pediatrul fara experienta in reumatologie si prezinta un grad ridicat de incertitudine la nivel de raspuns terapeutic, evolutie si prognostic.

DIAGNOSTIC AND THERAPEUTIC ISSUES IN SYSTEMIC VASCULITIS IN CHILDREN

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Paediatric systemic vasculitis is a group of disease with low incidence, with heterogeneous clinical symptoms and raising difficult issues of diagnosis and treatment. The common element is the inflammation of the vascular wall, some forms also associating extravascular necrotizing granuloma. The clinical manifestations are both the general symptoms and the symptoms specific to the affected organs.
The authors discuss the changes occurred in nomenclature, the classification and diagnostic criteria of international consensus, starting from Chapel-Hill (1994 and 2012) and continuing with the criteria proposed by EULAR / PRINTO / PRES in 2010, the latter being the result of an international group recognized as the European leader in paediatric rheumatology. The reunited EULAR / PRINTO / PRES groups validated the diagnostic criteria for the four main paediatric vasculitis: Henoch-Schönlein purpura, Polyarteritis child nodosa, Wegener's granulomatosis and Takayasu's arteritis. Here, it is summarized and the score of vasculitis activity evaluation in child - PVAS (vasculitis Paediatric Activity Score), and the therapeutic options available today.
The authors point out that despite diagnostic and therapeutic progress registered in the last two decades, systemic vasculitis in children continue to have a high degree of poor diagnosing by a paediatrician with no experience in rheumatology and shows a high degree of uncertainty at therapeutic response, evolution and prognosis.

URGENŢE – TOXICOLOGIE / PEDIATRIC EMERGENCIAS – CLINIC TOXICOLOGY

54. URGENŢELE HIPERTENSIVE ÎN INTOXICAŢIILE ACUTE LA COPIL ŞI ADOLESCENT

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Urgenţele hipertensive sunt acele situaţii care necesită scăderea promptă a valorilor tensiunii arteriale pentru a preveni sau a reduce riscul afectării organelor ţintă. Hipertensiunea arterială secundară poate face parte din tabloul clinic al intoxicaţiilor acute cu medicamente din diferite clase farmacologice cardiovasculare şi noncardiovasculare, substanţe de abuz, substanţe chimice sau porţiuni de plante. Monitorizarea semnelor vitale atât la prezentare, cât şi în dinamică este esenţială în abordarea acestor cazuri. Cel mai adesea, hipertensiunea arterială secundară intoxicaţiilor acute este uşoară şi tranzitorie. Deşi rare, atunci când apar în tabloul clinic al cazurilor de intoxicaţie acută la copil şi adolescent, urgenţele hipertensive reprezintă o condiţie ameninţătoare de viaţă. În absenţa unui diagnostic şi tratament prompt, acestea pot genera sechele grave. Pe de altă parte, o intervenţie terapeutică mult prea entuziastă poate duce în egală măsură la complicaţii.

HYPERTENSIVE EMERGENCIES IN ACUTE POISONING IN CHILDREN AND ADOLESCENTS

Coriolan Emil Ulmeanu, Viorela Gabriela Niţescu, Cristina Iolanda Vivisenco

Hypertensive emergencies are those situations that require immediate blood-pressure reduction in order to prevent or limit the risk of target organ damage. Secondary hypertension may be part of the clinical picture of acute poisoning with drugs from different cardiovascular and non-cardiovascular pharmacological classes, drugs of abuse, chemicals and plants. Vital signs monitoring, both at presentation and in evolution is essential in these cases. Most often, secondary hypertension in acute poisoning is mild and transient. Although infrequent, when they occur in the clinical picture of acute poisoning in children and adolescents, hypertensive emergencies represent a life-threatening condition. In the absence of prompt diagnosis and treatment, they can generate serious sequelae. On the other hand, an overly enthusiastic therapeutic intervention can equally lead to complications.

55. ROLUL SIMULARII IN PROCESUL EDUCATIONAL AL MEDICINEI DE URGENTA IN PEDIATRIE

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4. Student UMF Carol Davila

Scopul ultim al oricărei intervenții efectuate la un pacient aflat în situație amenințătoare de viață este prevenția decesului. Toate aceste manevre trebuie efectuate rapid și de către un personal medical și paramedical antrenat. Soluția pentru diseminarea cunoștințelor de resuscitare la un număr din ce în ce mai mare de potențiali salvatori este instruirea prin simulare. Simularea în medicina modernă a apărut ca urmare a faptului ca unele proceduri nu pot fi învățate de către student sau rezident prin aplicare directă la pacient, chiar și sub atenta și directa supraveghere a unui medic experimentat și calificat în a practica procedura respectivă. Această imposibilitate este rezultatul fie al unor considerente etice – riscurile pentru pacient și potențialele complicații sunt foarte mari în mâini neexperimentate – fie ale unora de natură tehnică – instrumentar foarte complicat, afecțiune foarte rară, procedură nou elaborată etc.

Grupul de lucru de la mânăstirea norvegiană Utstein în anul 1990 a standardizat terminologia si nomenclatura folosită în cercetarea si educația privind stopul cardio-respirator în afara spitalului. De aici au plecat strategiile de implementare ale tehnicilor de simulare, în paralel cu standardizarea intervențiilor salvatoare de viață [apariția ghidurilor de Basic și Advanced Life Support]. Aceste situații apar în peste 80% din cazuri acasă, în locuri publice, în locuri izolate. Adică în afara spitalului sau la distanță de un centru medical abilitat în soluționarea urgentă a cauzei amenințătoare de viață. Iar la copil lucrurile sunt și mai dificil de rezolvat datorită faptului că atât anatomic cât și funcțional și farmacologic, copilul nu este „un adult în miniatură”.

Simularea în medicina de urgență pediatrică a luat amploare ca urmare a standardizării tehnicilor de control avansat adresate căii aeriene, ale manevrelor de masaj cardiac, de acces vascular, de stimulare electrică, etc. Primele modele de simulare au fost aplicate la cadavru sau pe modele artificiale cu un grad redus de realism. Lipsa de veridicitate și lipsa unei standardizări a ritmicității reinstruirii au dus la ameliorări modeste ale ratei de mortalitate în incidentele și accidentele pediatrice. In ultimele decenii au apărut manechine din ce în ce mai performante și mai realiste [Laerdal, Ambu, Simulaids, etc].

Lucrarea de față se dorește o punere la punct pentru procedurile de simulare cel mai frecvent folosite în medicina de urgență pediatrică și o documentare a eficacității aplicării acestora intr-un spital universitar de mono specialitate [exclusiv pediatric] pe durata a peste 20 de ani de când există echipa acreditată de instructori PALS la IOMC Alfred Rusescu.

SIMULATION IN EMERGENCY PEDIATRICS TRAINING PROCESS

The ultimate purpose of a medical intervention in a critical situation is death prevention. All procedures should be performed by a skillful medical team. Dissemination of knowledge to a large number of potential savers is by scenarios and simulation.

Procedures in emergency medicine cannot be performed in real case scenarios by training doctors, even with direct supervision of an experienced tutor, because of ethical reasons or technical issues.

Since 1990 when Utstein Abbey group generated a set of precise definitions and terminology for out-of-hospital cardiac arrest, research in emergency medicine and resuscitation guidelines were generated and implemented. 80% of arrest cases occur at home or in a public place, not in a hospital or a medical facility. In children such emergencies are even more difficult because of anatomical and functional particular aspects. A child is not “a miniature adult”.

Simulation in pediatric emergency medicine has evolved since BLS and PALS guidelines were implemented and advanced control measures for airway, chest compression and defibrillation, vascular access were standardized. Initial scenario training was cadaveric or on non-realistic models, with little improvement in resuscitation outcome. In the last decades very realistic models were developed – Laerdal, Ambu or Simulaids manikins are available.

This presentation is a review of simulation areas available in pediatric emergency medicine and is documented with the significant experience [more than 20 years] of the training group from IOMC Alfred Rusescu.

56. SEPSISUL SEVER SI SOCUL SEPTIC LA COPIL - UP TO DATE

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Sepsisul sever reprezinta in continuare o cauza majora de morbiditate si mortalitate nu numai in tara noastra, ci si in intreaga lume. Sepsisul la copil este sub-recunoscut si deseori este diagnosticat tardiv si tratat necorespunzator datorita confuziei simtomatologiei, a necunoasterii definițiilor de sepsis, precum si datorita aplicarii inconsecvente a ghidurilor clinice standardizate. Incidenta sepsisului este în crestere alarmanta in ciuda progreselor privind fiziopatologia bolii precum si a posibilitatilor terapeutice actuale din terapiile intensive moderne. Potrivit Organizatiei Mondiale a Sanatatii, sepsisul provoaca intre 60-80% decese pe an in copilarie. Ghidurile si initiativele recente au aratat ca progresia de la sepsis la sepsis sever sau soc septic poate fi oprita si inversata prin interventii terapeutice timp-sensibile. Astfel, în acest review vom discuta

definitiile standardizate ale sepsisului, evidentele acutuale privind resuscitarea cu fluide, suportul vasoactiv, antibioterapia precum si alte masuri generale de sustinere in sepsis. Scopul nostru general este acela de a furniza clinicienilor o abordare sistematica actualizata pentru a trata prompt si corect sepsisul la copii.

SEVERE SEPSIS AND SEPTIC SHOCK – UP TO DATE

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Severe sepsis remains a major cause of morbidity and mortality not only in our country, but around the world. Sepsis in children is under-recognized and is often inadequately treated due to late diagnosis, confusion of clinical symptoms, lack of knowledge about the definitions of sepsis and inconsistent application of standardized clinical guidelines. The incidence of sepsis is increasing dramatically despite improved understanding of the pathophysiology and therapeutic possibilities existing in modern intensive care units. According to the World Health Organization, sepsis accounts for 60-80% of lost lives per year in childhood. Recent guidelines and initiatives have shown that progression from sepsis to severe sepsis or septic shock can be stopped and reversed by time-sensitive therapeutic interventions. Thus, in this review we will discuss standardized definitions of sepsis, the evidence of resuscitation fluids, vasoactive support, antibiotics and other general supportive measures in sepsis. Our overall goal is to provide clinicians an updated systematic approach to treat sepsis in children in a quick and accurate manner.

57. GHIDURI DE TRATAMENT IN BRONSIOLITA ACUTA – PLUSURI SI MINUSURI

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2 – Spitalul Clinic de Urgenta pentru Copii ”Louis Turcanu” Timisoara

Bronsiolita acuta reprezinta una dintre cele mai frecvente afectiuni la sugar si copilul mic. In ciuda numarului mare de studii efectuate pe aceasta patologie nu exista in prezent un consens privind cea mai buna atitudine terapeutica.
De-a lungul timpului trei principale resurse terapeutice si-au disputat intaietatea si anume: salbutamol/albuterol (administrat in aerosoli sau sub forma de pufuri), adrenalina/epinefrina si serul hiperton sub forma de aerosoli. Conform recomandarilor Academiei Americane de Pediatrie din 2014 nici una dintre aceste resurse terapeutice nu este eficienta in scurtarea duratei de evolutie a bolii iar raportul beneficiu/risc inclina semnificativ in favoarea riscului (Recomandari de tip B). De asemenea ghidul contraindica administrarea de corticosteroizi in bronsiolita (Recomandare de tip A) si efectuarea de fizioterapie respiratorie (Recomandare de tip B). Administrarea de rutina de antibiotice si antivirale nu este de asemena indicata (Recomandare de tip B).
Lucrarea prezinta o analiza critica a studiilor care au stat la baza acestor recomandari, in paralel cu un studiu retrospectiv efectuat pe 236 de pacienti cu bronsiolita internati in Clinica I Pediatrie Timisoara pe o durata de un an.

TREATMENT GUIDELINES IN ACUTE BRONCHIOLITIS - PLUSES AND MINUSES

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2 – ”Louis Turcanu” Emergency Hospital For Children Timisoara

Acute Bronchiolitis is one of the most common diseases in infants and young children. Despite the large number of studies on this pathology there is currently no consensus on the best therapeutic approach. Over time three main therapeutic resources have played primacy: salbutamol/albuterol (administered in the form of aerosols or sprays), adrenaline/epinephrine and hypertonic saline as aerosols. According to the recommendations of the American Academy of Pediatrics developed in 2014 none of these therapeutic resources is effective in shortening the duration of the disease and the benefit/risk ratio significantly tilts in favor of risk (Evidence Quality B). The guideline also contraindicate the administration of systemic corticosteroids in bronchiolitis (Evidence Quality A) and respiratory physiotherapy performance (Evidence Quality B). Routine administration of antibiotics and antivirals is also not indicated (Evidence Quality B). This paper presents a critical review of the studies that were the basis of these recommendations, along with a retrospective study on 236 patients hospitalized with bronchiolitis in First Pediatric Clinic Timisoara for a period of one year.

SIMPOZIOANE SATELIT / SATELLITE SYMPOSIUMS

SESIUNE COMUNĂ SOCIETATEA ROMÂNĂ DE PEDIATRIE ŞI SOCIETATEA TURCA DE PEDIATRIE
THE ROMANIAN AND THE TURKISH SOCIETIES OF PEDIATRICS JOINT SESSION

1. ASPECTS OF PEDIATRIC CARE IN TURKEY AND DEMOGRAPHIC PHENOMENA

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Summary: Turkey has a large population with almost 30 % of it under 18 years old and has achieved important improvements on health system recently. Some characteristics of the country and health status are given and current pediatrics education system are summarised in this manuscript.

Turkey, known officially as the Republic of Turkey, is a Eurasian country that stretches across the Western Asia and southeastern Europe. Turkey has a large population with almost 30 % of it under 18 years old.

In 2013 the life expectancy in TURKEY increased to 77.0 years. That year, the life expectancy for women was 79.40 years and for men 73.70 years. Turkey's position is 37th in 180 countries. Infant mortality rate per 1000 live births reached 7,7 in 2012. Our babies are widely protected from phenylketonuria, congenital hypothyroidism, biotinidase deficiency and cystic fibrosis by country wide neonatal screening program. Free of charge iron and vitamin D support to babies were provided.

Many important achivements have been successed in health system recently. National Tobacco Control Program, Measles Eradication Program and Baby Friendly Hospitals Initiative are some of thems. Smoking is an important public health problem in our country. Our country ranked in the 7rd place in the world in terms of tobacco consumption. The "Tobacco Control Framework Agreement" prepared by WHO was signed in 2004, and the "National Tobacco Control Program" was declared in 2007. There has been a great support to the measures which implementation started in 2008 to prevent passive smoking and Turkish smokers declined 13.4 percent since 2008.

Effective combat against communicable diseases such as tuberculosis, malaria and measles was carried out. A wide vaccination campaign for eradication of measles, covering the years 2003-2005 had been conducted and 18,217,000 children were vaccinated within the campaign which has been the widest campaign ever done in the Turkey and Europe. The vaccination rate was % 97. Number of the measles cases was 30,509 in 2001 and it decreased to 4 in 2008. Vaccination campaign was widened to cover Syrian refugees in 2013. The vaccination rate for other infections is over % 95.

Baby Friendly Hospitals Program are still carrying out and the number of "Baby Friendly Hospitals reached 1007 at the end of 2013. We are continuing "Promotion of Breastfeeding and as a result, the ratio of exclusively breast feeding between 0-6 months of age is increased to 40.4 % in 2009. Additionally, all obstetric units have staff who certified as attending "Newborn Resuscitation Training Program".

History of medical education in Turkey

The history of medical education can be evaluated in three parts as follows;

1. Before 1970-Traditional (Flexnerian) period: Following foundation of Istanbul University Medical School (1909), as a wise policy, the young Republic of Turkey, tried to increase number of physicians and tertiary hospitals across the country. Prof. Dr. Dogramacı had founded Hacettepe University Medical School which had an innovative integrated curriculum adapted from United States of America' University model.
2. 1970-1996-Quantity movement: Under pressure of rapid changes in demographic and socio-cultural figures at the country, such as high increase of the population, urbanization, increase of social demands, modernization desires, Turkish governments used "increasing health manpower" policy. During the late 1980s and early 1990s the number of medical schools is increased as well as the number of students accepted to these schools.
3. 1996-2014 Quality movement: This period is characterized by stable student number and several initiatives and innovations.

Today, 82 medical schools are accepting nearly 11000 new students and educating 32,807 undergraduate students. Now we are the 5th country respect to number of the medical schools in the world and first in Europe. Number of student per lecturer in faculties of medicine is 3,5 student/lecturer in Turkey (Europe:3,3 and USA:2,5 student/lecturer).

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2. REFLUXUL GASTROESOFAGIAN LA NOU-NĂSCUT ŞI SUGAR

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Refluxul gastroesofagian (RGE) este o entitate frecventă în pediatrie, care este încă subdiagnosticată (”boala mileniului trei”). Simptomele clinice polimorfe sunt dominate nu numai de vărsături post-alimentare, ci în multe cazuri şi de simptome respiratorii (de la tuse nocturnă la episoade de apnee, wheezing recurent, pneumonie de aspiraţie şi chiar sindromul de moarte subită). Terapia RGE este triplă: măsuri generale, terapie farmacologică şi chirurgicală. Drogurile iniţiale (agenţi prokinetici) au fost înlocuite în ultima perioadă cu inhibitorii pompei de protoni (IPP), deoarece acestea sunt considerate medicamente în măsură să determine dispariţia simptomelor de reflux în cele mai multe cazuri. O abordare corectă a tratamentului şi o identificare precoce a bolii previne apariţia complicaţiilor şi o calitate mai bună a vieţii.

GASTROESOPHAGEAL REFLUX IN NEW-BORN AND INFANTS

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Gastroesophageal reflux (GER) is a frequent entity in the pediatrics that is still underdiagnosed ("the third millennium's disease"). Clinical polymorphic symptoms are not only dominated by post-food vomiting, but in many cases and respiratory symptoms (night coughing at episodes of apnea, recurrent wheezing, aspiration pneumonia and even sudden death syndrome). GER has triple therapy: general measures, surgical and pharmacological therapy. The initial drugs (prokinetics agents) were replaced in the last period with proton pump inhibitors (PPI), because they are considered drugs able to cause the disappearance of the symptoms of reflux in most cases. A correct approach of the treatment and early identification of the disease prevent the complications occurrence and a better quality of life.

SIMPOZION INTERNAŢIONAL - SATELIT
INTERNATIONAL SATELLITE SYMPOSIUM

3. ADVANTAGES OF PREVENTIVE NURSE SYSTEM

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Background
The infant mortality rate was 26.4 % in Hungary in 1915, the highest in Europe. Certain Nations and the European Union have initiated numerous programme for turning the natural decrease in Europe.
Objective
All child has right to adequate care and supply. A Perventive/visiting nurse system was established in 1915 in Hungary. Preventive nurse mostly deals with the families in two developmental period. During the pregnancy and when the child is between 0-7 years old. Preventive nurse cares the families and completes family doctors work.
Results
They perform different examinations independently. They deal with the pregnant, organize preventive screening for them. The first home visit of newborn after delivery is compulsory at first 48 hours for them. They control the newborns, the infants during the first year of age frequently, and little bit rare in childhood at home and in an office. The infant mortality rate decreased to 5‰ in 2014. The vaccination rate 99.9%. The exclusive breasthfeeding 50% under 6 month as the result of preventive nurse system.
Conlcusion
We organize an 8 semester BSc course for preventive nurse in English at Semmelweis University, Budapest

4. URINARY SCREENING IN 6-12YEARS SCHOOL CHILDREN IN SAUDI ARABIA
“ Makkah and Al-Baha region”

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*KFH, Al Baha and ** Oum Alkurra University Hospital

Purpose:
Is to screen school children in Makkah and Al-Baha region in Saudi Arabia, for prevalence of hematuria , proteinuria and hypercalciuria, the two regions represent two different population in term of consanguinity rate and ethnicity.
Methods:
This cross sectional study, approved by the Institutional Review Board Committees of Makkah and Al Baha, was carried out between October 2012 and April 2013 on 12352 children (including 9299 children from Makkah and 5053 children from Al Baha).

The urine samples were collected in sterile containers, from children in selected elementary schools from different districts in Makkah and Al-Baha. Proteinuria, hematuria or hypercalciuria were looked for by urine Dip Stick.

Results:

In Makkah region prevalence of hematuria was 11.6% and 4.7%, in male and female, respectively, for proteinuria prevalence was 3.1% and 2% in male and female respectively. The prevalence of hypercalciuria was 10.4% and 6.1% in male and female respectively, none of hematuria cases were hypercalciuric. In Al Baha region prevalence of hematuria was 24.5% and 10.9%, in male and female, respectively, for proteinuria prevalence was 3% and 1.6% in male and female respectively. The prevalence of hypercalciuria was 20.8% and 8.6% in male and female respectively, and 67.6% of male hematuria cases has hypercalciuria, and 71.5% of female hematuria cases has hypercalciuria.

Conclusion:

So, asymptomatic urinary abnormalities might be detected by urine screening program at school age. Further work-up should be offered to define the exact etiology of any abnormal finding and to determine whether early detection of renal disorders in childhood will lead to effective interventions and reduction in the number of individuals who develop end-stage renal disease.

5. BREAST FEEDING PRACTICES IN AL-BAHA REGION, SAUDI ARABIA FACTORS AFFECTING I

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Purpose:

Although the incidence of B.F. in Saudi Arabia has been declining over the past two decades, the reasons behind this decline have not been investigated. To the best of our knowledge, there are no statistics available regarding B.F. at Al-Baha level. This information is crucial for the development of a successful B.F. promotion plan in this local community. The objective of this study was to find out the prevalence of B.F., the social, demographic and medical influencing factors related to the initiation and continuation of B.F., the knowledge and attitude of females towards B.F. in Al-Baha, Saudi Arabia. Also, to determine the weak points and common misbeliefs, which should be considered during the intervention stage.

Methods:

The study was conducted in Al-Baha region, Saudi Arabia in the period between January and November 2013. The participants were 714 mothers having 714 infants below 24 months, from both sexes and equally distributed among six age groups. All the sampled mothers completed answering questionnaires consisting of personal information, data regarding the common factors affecting breast feeding and the current breast feeding practice.

Results:

The results showed that the current overall B.F rate was only 22.4%, current exclusive B.F rate was only 3.1%, partial B.F rate was 19.3% and bottle feeding rate was 77.6%, 28.6% of the sampled mothers continued B.F for one year and only 9.2% of them continued B.F (currently) for two years. The factors most significantly associated with the outcome of B.F were the misperceptions about B.F, mother's education, advice received regarding B.F, antenatal care, mother's work status, mode of delivery and milk samples given on discharge from hospital after labour. The mother's knowledge about breast feeding was very poor.

Conclusion:

The current study emphasizes that the most significant factors affecting the outcome of B.F are highly modifiable by health education. This information is crucial for the development of a successful B.F promotion plan in this community. All the local hospitals should be prepared to be certified as baby friendly hospitals.

6. SURFACTANT IN PAEDIATRIC PATIENTS WITH ARDS

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Acute respiratory failure remains the primary indication for admission to paediatric intensive care units (PICUs) and accounts for significant mortality, morbidity and resource utilization. Respiratory infections, in particular pneumonia and severe bronchiolitis, are the most common causes of respiratory failure requiring mechanical ventilation in children.

Alterations in endogenous surfactant play a role in the pathogenesis of many causes of acute lung injury (ALI) and acute respiratory distress syndrome (ARDS). Surfactant dysfunction, destruction and inactivation have also been demonstrated in children with acute respiratory insufficiency due to bronchiolitis. The administration of exogenous surfactant may reduce the need for mechanical ventilation and its associated sequelae by restoring surfactant levels and function. Investigators have studied exogenous surfactant in other populations with various causes of respiratory failure. In children with respiratory failure, the efficacy of exogenous surfactant has been suggested in uncontrolled studies. The relatively low mortality rate, the diversity of the study populations and the shorter duration of mechanical ventilation are factors that make large-scale randomized controlled trials in this population challenging to conduct. While the use of surfactant in ARDS/ALI has not been previously systematically reviewed, its use in children with bronchiolitis has been (1).

Surfactant use decreases mortality, is associated with more ventilator-free days and reduces the duration of ventilation. No serious adverse events have beed reported so far It seems the bestter result is in patients with the pulmonary origin of ARDS. Most trials enrolled small numbers of children, and further well-designed and adequately powered multicentre trials are therefore required.

An overview of children in PICU in University Medical Centre Ljubljana (Slovenia) who were treated by surfactant is presented. In three years the surfactant therapy was administred in only 11 non-neonatal patients.

Ref.: 1. Duffett M , Choong K, Ng V et als: Surfactant therapy for acute respiratory failure in children: a systematic review and meta-analysis. Critical Care 2007,11:R66.

SIMPOZION ROMANO - ITALIAN SATELIT
ROMANIAN - ITALIAN SATELLITE SYMPOSIUM

7. CE TREBUIE STIUT DESPRE COPILUL CARE SFORAIE

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Sforaitul este un simptom comun in randul populatiei pediatrice care apare la copii in toate decadele de varsta, avand o incidenta crescuta in perioada de prescolar (10%). Acest zgomot poate fi declansat de mai multi factori (*alergii sezoniere, infectii de cai respiratorii, deviatie de sept nazal, apnee obstructiva de somn, etc*).

Calitatea somnului influenteaza viata cotidiana a fiecarui copil, avand un rasunet important asupra viitoarei dezvoltari cognitive si comportamentale a acestuia. Cercetarile intreprinse in domeniu incearca sa stabileasca o legatura intre comportamentul zilnic al copilului, comorbiditatile asociate si tulburarile respiratorii din timpul somnului secundare sforaitului.

Sforaitul este un sunet de intensitate variabila, produs in somn, caracterizat ca deranjant, emis in timpul actului respirator atunci cand fluxul de aer este partial blocat. Ca o consecinta a limitarii fluxului de aer se produce vibratia tesuturilor din jurul cailor respiratorii superioare, putand asocia tulburari respiratorii de somn.

Tulburarile respiratorii asociate somnului descriu un spectru larg de entitati fiziopatologice secundare obstructiei partiale sau complete a cailor respiratorii superioare in timpul somnului, de la sforaitul primar la sindromul de rezistenta al cailor respiratorii superioare sau apneea obstructiva de somn.

Sforaitul este un proces complex, rezultat al interrelatiei dintre posibile anomalii ale cailor respiratorii ca urmare a unor aspectele particulare cranio-faciale (*structura cranio-faciala, hipetrofie adenoido-amigdaliana, obezitate*, etc), asociat cu un control neuromuscular inadecvat (activitatea reflexa de dilatare ca raspuns la obstructia cailor respiratorii este anormala) la care se pot asocia si alti factori posibili (*boli genetice, boli de metabolism, boli infectioase, fumat, etc*).

Calitatea somnului influenteaza viata cotidiana a fiecarui copil, astfel incat prezenta sforaitului, fragmentarea somnului, perioadele de somnolenta excesiva in timpul zilei impun screening si evaluare medicala complexa din partea corpului medical (pediatru, ORL-ist, neurolog, pneumolog, psihiatru, etc). Istoricul clinic al pacientului nu este intotdeauna un indicator fidel al gradului de obstructie al cailor respiratorii superioare si al prezentei sau al severitatii tulburarilor respiratorii asociate somnului, astfel incat medicul trebuie sa acorde o importanta deosebita tuturor examenelor clinice de rutina.

In lipsa unui tratament, sforaitul poate fi elementul care sta la baza dezvoltatii, in dinamica, a unor co-morbiditati cardiovasculare, neurocognitive, comportamentale, somatice sau metabolice. Anxietatea, deficitul de atentie, depresia, imposibilitatea integrarii sociale ajung sa devina stigmate zilnice ale copiilor care sforaie. Copiii cu tulburari respiratorii asociate somnului au sanse mai mari sa dezvolte sindrom metabolic, comparativ cu copiii fara tulburari respiratorii asociate somnului. De asemenea, exista studii care sustin ideea ca fragmentarea somnului poate favoriza cresterea si dezvoltarea tumorala.

Polisomnografia este considerate metoda gold-standard in evaluarea copilului care sforaie, iar adenoidotonsilectomia este indicatia chirurgicala de electie in tulburarile respiratorii asociate somnului la copil.

Abordarea unui copil care sforaie este complexa si presupune respectarea unor strategii de educare a somnului, folosirea dispozitivelor anti-sforait, terapie medicamentoasa sau interventie chirurgicala.

Variabilitatea factorilor de risc implicati in modelarea comportamentului zilnic si a dezvoltatii neurocognitive a copilului precum si in aparitia complicatiilor, determina perfectionarea si evaluarea continua a metodelor de diagnostic si tratament.

Cuvinte cheie: sforait, copil, tulburari respiratorii asociate somnului, sforait primar, afectiuni organice.

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WHAT IT IS TO BE KNOWN ABOUT A SNORING CHILD?

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Snoring is a common symptom in paediatric population, occurring in children of all ages but being more commonly seen in preschool-aged children(10%). Soring can be associated or determined by many factors (*allergies, respiratory tract infections, nasal deviation, obstructive sleep apnea, etc).*

A good night's sleep influences every days' quality of life, having a boomerang effect on the child's future cognitive and behavioural development. Researches in this field try to establish a connection between a child's daily behaviour and sleep-related breathing disorders due to snoring.

Snoring is an annoying noise of variable intensity, produced during breathing and while sleeping, secondary to partial obstruction of the upper airways. As a consequence of this restricted airflow, vibrations appear within the upper airway's soft tissue and this may lead to sleep-related breathing disorders.

Sleep-related breathing disorders refer to a large spectrum of pathophysiologic entities that appear secondary to partial or total obstruction of the upper airways, from primary snoring to upper airway resistance syndrome and obstructive sleep apnea.

Snoring is a complex process, the result of interaction between possible upper airway anomalies due to particular cranio-facial anatomy (cranio-facial structures, adenoidal hipertrophy, obesity, etc) and abnormal neuromuscular control (a dilatation reflex activity in response to upper respiratory obstruction often fail) or any other factors (genetic diseases, metabolic diseases, infectious diseases, smoking, etc).

A good night's sleep influences every day quality of children's life and this is why snoring, fragmented sleep, daytime excessive sleeping need screening and a careful medical examination from medical specialists (paediatrician, ENT, neurologist, pneumologist, psychiatrist, etc). A patient's clinical history is not always a good way to establish the degree of the upper airway obstruction or the severity of sleep-related breathing disorders, this is why the doctor needs to pay attention to every clinical routine examination.

Without treatment, snoring can become the pathway for developing cardiovascular co-morbidities, neurological and behavioral pathology, somatic or metabolic diseases. Anxiety, attention problems, depression, social impairment become daily routine for a snoring child. Children with sleep-related breathing disorders have more chances in developing a metabolic syndrome compared with children without sleep-related breathing disorders. Also, there are studies that support the idea that fragmented sleep accelerates cancer growth and tumor progression.

Polisomnography is the gold-standard in evaluation of a snoring child and adenotonsillectomy is the first-line treatment for children with sleep-related disorders breathing.

The management of a snoring child is a complex process that includes educational strategies for a good sleep hygienne, use of anti-snoring devices, medication and surgery.

The variability of risk factors involved in the daily modelling of a child's behaviour and his neurocognitive development, as well as the ones involved in the complications rate, determines the perfecting progress and continuous evaluation of diagnostic and treatment methods.

Key-words : snoring, child, sleep-related breathing disorders, primar snoring.

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8. METABOLOMICS IN PERINATAL MEDICINE: EVOLUTION OR REVOLUTION?

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Introduction. The primary target of neonatology is to guarantee the healthcare of newborns and use all the available resources to increase the survival rate of those neonates who face critical pathological conditions. Over time, in line with this necessity, different areas of scientific research emerged, with a focus on better understanding the pathogenic mechanisms of diseases which occur in clinical practice. Recently an important scientific contribution comes from metabolomics, defined by Nicholson as the quantitative measurement of the dynamic multiparametric metabolic response of living systems to pathophysiological stimuli or genetic modifications. The delineation of metabolic response is possible by the analysis and measurement of the metabolites that the organism produces to response at external stimuli or internal message.

Research lines. The key role of metabolomics in neonatology emerges in the studies present in literature which analyze the most common diseases in clinical practice such as neonatal sepsis, perinatal asphyxia and intrauterine growth restriction (IUGR).

Neonatal sepsis

Sepsis is a clinical syndrome characterized by a series of immunological, metabolic, haemodynamic and respiratory alterations secondary to an infective process caused by an abnormal systemic inflammatory response syndrome in the organism. Septicemia and infections disease are the first causes of death in neonatal period (32%). Despite several are the biochemical parameters used as diagnostic tool in clinical practice, there are still limits about sensitivity and specificity in the use of this markers. Once again the purpose of recent -omic science is to reach higher standard in early diagnosis, or in the individuation of population at risk. The identification of metabolomic trajectories which could assume the neonatal organism during sepsis was taken as object of analysis by Fanos et al. Using a combined approach based both on 1H-NMR and gas-chromatography-mass spectrometry (GC-MS), in addition with multivariate statistical analysis, were studied 25 sample of urine: 9 from septic patients, and 16 from healthy controls. This study showed a unique metabolic profile of the patients affected by sepsis compared to non-affected ones with a statistically significant difference between the two groups. The metabolites that allow the different pattern between cases and controls were the major concentrations in urine of septic patients of lactose, maltose and glucose.

Perinatal asphyxia

Perinatal asphyxia is a pathological condition attributed to the hypoxia and/or ischemia event around the time of birth. This pathological state affects 4 million neonates worldwide per year, causing the decease of 1 million subjects. So far, there are new emerging theories in which is demonstrated the importance of the metabolomics approach in identifying the metabolic fingerprint of hypoxia/asphyxia conditions in term of metabolic fingerprint. A metabolomics study was performed by Longini et al, who investigated the effects of perinatal asphyxia on urine collected from 6 newborn patients and 8 healthy controls. Authors used a 1H-NMR approach with the aim to have a global low molecular weight metabolic profile of the samples. The analysis highlighted the main metabolic differences between the two groups. In particular the metabolites resulted characterizing the asphyxiated group sorted by importance were lactate, glucose and TMAO, dimethylglycine, dimethylamine, TMAO, creatine, glucose, succinate, threonine and 3-hydroxyisovalerate, formate, urea and aconitate.

IUGR

With the term of Intrauterine Growth Retardation are indicates the fetus who fails to reach its potential growth, with birth-weight below the 10th centile for their gestational age. This condition affected from 4 to 8 % of neonates born in industrialized country, while the incidence increased in the developing country, where IUGR rate in newborns population is about 6-30%. Dessì et al. (2011) were the first research group who had studied the metabolomic profile of IUGR in infants.

This study was based on urine samples collected from 56 infants that 26 were IUGR newborns and 30 belong to control group. Thanks to the use of 1HNMR spectroscopy, it was possible to discriminate the metabolites differences in the urine of cases and controls. In particular the authors found an increase of myoinositol concentration in IUGR patients. This metabolite has often been associated with glucose intolerance and insulin resistance in adults, and in near future could become also a valid marker of altered glucose metabolism during fetal development in IUGR.
Conclusions. Each of these pathologies is a daily challenge for neonatologist. Thanks to metabolomics is possible to obtain better knowledge of these problems in order to improve the life expectancy of newborns.
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9. MANIFESTĂRI RESPIRATORII ÎN BOALA DE REFLUX GASTROESOFAGIAN LA COPIL

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Boala de reflux gastroesofagian (BRGE) reprezintă trecerea intermitentă sau permanentă a conţinutului gastric în esofag. Datorită simptomatologiei heterogene, refluxul gastroesofagian prezintă destule capcane de diagnostic îndeosebi la sugarul mare, copil şi adolescent, unde regurgitaţiile şi vărsăturile, tipice sugarului mic, sunt înlocuite de manifestări polimorfe respiratorii şi neurocomportamentale. Managementul actual al BRGE îl reprezintă controlul simptomatologiei şi al impactului său major asupra calităţii vieţii. Un reflux necunoscut, netratat, sau insuficient tratat, care evoluează pe o anumită perioadă de timp, poate avea consecinţe digestive, respiratorii, neurologice, nutriţionale. În cazul asocierii BRGE cu manifestări respiratorii, terapia cu IPP este controversată, literatura de specialitate rămânând a fi revizuită pentru a pune în lumină această boală complexă.
Cuvinte cheie: boala de reflux gastroesofagian, manifestări respiatorii, copil

RESPIRATORY SYMPTOMS IN GASTROESOPHAGEAL REFLUX DISEASE IN CHILDREN

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Gastroesophageal reflux disease (GERD) consists of intermittent or permanent passage of the gastric content into the esophagus. Due to the heterogeneous symptomatology, the gastroesophageal reflux has enough diagnosis catches, especially in the older infant, child and teenager, where the vomiting and regurgitations of the infant are replaced by polymorphic symptoms, respiratory and neurobehavioral. The actual management of the GERD is focused on the control of the symptoms and on the major impact of the disease over the quality of life. If respiratory symptoms are associated with GERD, the PPI therapy is controversial, literature remaining to be reviewed in order to shed light on this complex disease.
Key words: gastroesophageal reflux disease, respiratory manifestations, child

10. THE EMERGING DISEASE: VIRUS EBOLA

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The viral desease caused by filo virus .In middle of last years this infection always they are becaming frequent and for sottuvalutation the problem and for the delays of help international intervent there has been at one drammatic development of this epidemy and just with one appropriated training of medics and with a prevent and manage is necessary a good information for people.

11. CORELATIA GENOTIP-FENOTIP IN MUCOVISCIDOZA (FIBROZA CHISTICA) IN PRACTICA CLINICA

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Mucoviscidoza (MV) sau fibroza chistică este cea mai frecventă anomalie monogenică a populaţiilor caucaziene, autosomal recesivă, cu evoluţie cronică, potenţial letală. Descoperirea genei patologice şi caracterizarea proteinei determinate, în anul 1989 a condus la aprofundarera informaţiilor privind mecanismele moleculare patogenice şi fiziopatologice implicate. Cercetarile ulterioare au demonstrat complexitatea problemei: gena MV este o genă mare, cu multiple alele patologice; identificarea mai multor mecanisme patogenice moleculare, demonstarea parţială a unor corelaţii semnificative genotip-fenotip; cunoaşterea insuficientă a celulelor ţintă (pentru terapia genică). Un proiect nou CFTR2 (Clinical an Functional Translation of CFTR) vizează o evaluare globala a funcţiei alelelor implicate din perspectiva explicarii relaţei genotip-fenotip, pentru ca la ora actuala este clar că nu doar funcţia pancreatica este corelata cu genotipul, respectiv, nu toate alelele descrise până în acest moment sunt importante pentru expresia fenotipica. Prezentarea aduce în discuție modul în care ne putem folosi în practica clinică de rezultatul testului genetic. Se prezinta cele 4 grupe "funcționale" de alele, care se pot depista în urma testului genetic molecular: alele patologice, alelele implicate în CFTR related disorders (bronșiectazii primare diseminate, pancreatita cronica/recurentă, absenta bilaterala a canalelor deferente - congenital bilateral absence of the vas deferens), alele fără consecințe clinice, respectiv cu funcție încă neelucidată.
Concluzii: Cazurile clasice de mucoviscidoza sunt de departe cele mai frecvente. Situatiile “particulare” trebuie discutate intre clinician si specialistul in genetică si tehnologia de diagnostic molecular. Diferenta intre formele fruste/ moderate de boala si CFTR related disorders nu este pe deplin clarificata. La cazurile incerte, genotipul trebuie sa contina cel putin o alela CFTR pentru a motiva investigatii ulterioare care sa dovedeasca diagnosticul sau cel putin sa motiveze necesitatea unei supravegheri speciale.

GENOTYPE-FENOTYPE CORRELATIONS IN CYSTIC FIBROSIS IN CLINICAL PRACTICE

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Cystic fibrosis (CF) is the most common monogenic autosomal recessive abnormality in Caucasian populations. CF has a chronic evolution and is potentially lethal. The discovery of the pathological gene and the characterization of the defective protein in 1989 led to a profound understanding of its pathogenic molecular and physiopathological mechanisms. Subsequent research demonstrated the complexity of the problem: the CF gene is a large gene with multiple pathological alleles; identifying multiple molecular pathogenic mechanisms, partial demonstration of genotype-phenotype correlations; insufficient knowledge of target cells (for gene therapy). A new project, CFTR2 (Clinical Translation of Functional year CFTR) seeks an overall assessment of the involved alleles’ function by explaining the genotype-phenotype relationship, because at this moment it is clear that not only the pancreatic function is correlated with the genotype, and not all the alleles described so far are important for the phenotypic expression. The presentation discusses how we can use the genetic test results in clinical practice. It shows the 4 groups "functional" alleles, which can be detected after molecular genetic testing: pathological alleles, alleles involved in CFTR related disorders (primary disseminated bronchiectasis, chronic/recurrent pancreatitis, congenital bilateral absence of the vas deferens), alleles without clinical consequences, with still unsolved function.
Conclusions: classic cystic fibrosis cases are by far the most common. "Uncommon" cases should be discussed between the clinician and the expert in genetics and molecular diagnostic technology. The difference between subtle/moderate forms of disease and CFTR Related Disorders is not fully clarified. In doubtful cases, genotype must contain at least one CFTR allele in order to justify further investigation for proving the diagnosis or at least to justify the need for special monitoring.

12. THE BULLOSA EPIDERMOLYSIS IN NEWBORNS: CLINICAL DIFFERENTIAL DIAGNOSIS
LA DIAGNOSI CLINICA E DIFFERENZIALE DELL’EPIDERMOLISI BOLLOSA IN EPOCA NEONATALE

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Definition: Hereditary epidermolysis bullosa is a group of genetically determined bullous diseases of the skin that share skin fragility as a common feature. This fragility is caused by mutations in various structural proteins of the epidermis and the dermoepidermal junction. The clinical symptoms and prognosis depend on which adhesive structure is missing, and on the location of the skin disruption. We describe four major types of EB, transmitted in AD or AR:

1. Simple Epidermolysis bullosa or epidermolytic
2. Junctional bullous epidermolysis

3. Dystrophic Bullous epidermolysis or Dermolytic BE
4. Kindler Syndrome

All the illness are characterized by the onset of blisters following minimal trauma than in normal subjects do not cause any injury.

Epidemiology The overall prevalence of simple EB, junctional EB, dystrophic EB in the population is estimated at 1/130.000 in the United States, 1/100.000 in Italy, 1/20.000 in Scotland. Kindler Syndrome is very rare, it is probably underdiagnosed, and currently about 200 cases are described.

Classification

Simple Epidermolysis bullosa or epidermolytic

Intraepidermal bullous are the most common lesions, representing the 50% of cases. The EBS are divided into two subtypes: basal E. due to cytolysis of basal keratinocytes, with the presence of predominantly cutaneous bullous lesions that resolve without scarring more; suprabasal E. where the lesions are formed in the suprabasal layers of the epidermis and include three different types extremely rare. They are transmitted in AD and AR form and are due to mutations in the genes KRT5, KRT14 (keratin 5 and 14), PLEC1 (plectin), PKP1 (Placofilin1), DSP (Desmoplachin).

Junctional bullous epidermolysis

Characterized by blisters between the epidermis and dermis at the level of the lamina lucida of the basement membrane. The lesions heal with the formation of hypertrophic tissue and/or atrophy. They originate from mutations transmitted in AR or AD. There are several variants due to mutation of genes LAMA3, LAMB3, LAMBC2 (laminin 3-3-2), COL17A1 (collagen type XVII), ITGB4 (integrine 4).

Dystrophic Bullous epidermolysis or Dermolytic BE

Bullous lesions are localized under the dense lamina of the skin basement membrane in the papillary dermis. They are characterized by slow healing with scarring and formation of milia. This form is about 25-35% of cases of EB. There are two major subtypes based on the mode of transmission AD or AR, with different clinical variants. All variants of Dystrophic EB are due to mutations in the COL7A1 gene coding for type VII collagen, the main component of the anchoring fibrils that ensure the adhesion of the basement membrane of stratified epithelia to the underlying mesenchyme.

Kindler Syndrome

Characterized by fragility of skin and mucosa, photosensitivity, progressive poikiloderma with extensive atrophy

Diagnosis Clinical features combined with immunofluorescence antigen mapping and/or electron microscopy examination of a skin biopsy allow to define the EB type and subtype. Molecular diagnosis is nowadays feasible in all EB subtypes and required for prenatal diagnosis. The extent of skin and mucosal lesions varies greatly depending on EB subtype and patient age. In the more severe EB subtypes lifelong generalized blistering, chronic ulcerations and scarring sequelae lead to multiorgan involvement, major morbidity and life-threatening complications. In the absence of a cure, patient management remains based on preventive measures, together with symptomatic treatment of cutaneous and extracutaneous manifestations and complications. Owing to its nature and severity, RDEB presents unique challenges for developing successful therapies that simultaneously alleviate the plethora of complications while having a significant impact on survival and quality of life. Recent approaches such as allogenic cellular therapy, gene therapy, and protein therapy show promise.

Differential diagnosis Congenital ichthyosis bullosa, incontinentia pigmenti, Staphylococcal pyoderma Bullous, Pemphigus luetic, autoimmune bullous diseases (pemphigus, herpes gestationis), Bullous mastocytosis, transient dermatolisis of newborn

LUCRARI PREZENTATE ORAL / ORAL PRESENTATION

CARDIOLOGIE / CARDIOLOGY

1. COARCTATIA DE AORTA LA SUGAR SI COPIL – DIAGNOSTIC, TRATAMENT SI PROGNOSTIC

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Scop: Dorim sa aducem in atentie coarctatia de aorta, cu cele doua moduri de exprimare clinica, la sugar si la copil. Prima forma reprezinta o urgenta, iar cea de a doua este de multe ori sub-diagnosticata, cu repercursiuni, prin prezenta si persistenta hipertensiunii arteriale la copii.

Material si metoda: Prezentam cateva cazuri de coarctatie de aorta, depistate la copiii care s-au adresat Clinicii III Pediatrie din Timisoara pentru hipertensiune arteriala, sindrom cefalalgic sau suflu cardiac. Toti au beneficiat de examen clinic, ECG, Rx cardiopulmonar, echocardiografie si selectiv, angio CT torace cu reconstructie 3D a cordului, pentru evidentierea zonei de coarctatie a aortei.

Rezultate: Am pus diagnosticul de corctatie de aorta, atat la sugari, cat si la copii. A fost prezenta hipertensiunea arteriala la membrele superioare, cu diferente de 20 mmHg intre membrele superioare si inferioare si absenta pulsului la artera femurala. Sugarii cu coarctatie de aorta au necesitat interventie chirurgicala urgent. Copiii au fost operati si ulterior monitorizati. A reaparut hipertensiunea arteriala si recoarctare in 2 cazuri, unul asociind arc aortic gotic. A fost necesara medicatie antihipertensiva si explorare interventionala cu montare stent intr-un caz si dilatare cu balonas in cazul cu arc aortic gotic.

Concluzii: Absenta pulsului la artera femurala in conditii de hipertensiune arteriala la membrele superioare pune diagnosticul de coarctatie de aorta. Postoperator pacientii trebuie monitorizati, datorita riscului de recoarctare. Dilatarea cu balonas interventional si montarea de stent sunt necesare in acest caz, dar pacientii trebuie monitorizati deoarece pot sa prezinte si ulterior hipertensiune arteriala restanta.

AORTIC COARCTATION IN INFANTS AND CHILDREN – DIAGNOSE, TREATMENT AND PROGNOSTIC

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Aim: To present aortic coarctation in infants and children, because infant coarctation is an emergency and child coarctation is sometime under-diagnosed, patients presenting at the hospital for high blood pressure of unknown etiology.

Material and methods: We want to highlight some cases of aortic coarctation admitted in the III-rd Pediatric Clinic Timisoara, for high blood pressure, headache or cardiac murmur. All of them performed clinical examination, ECG, Cardiopulmonary X ray, echocardiography and selective angio CT with 3D reconstruction.

Results: Aortic coarctation was diagnosed in infants and children. High blood pressure in the upper arms, with 20 mmHg difference between upper and lower arms was present, associated with absent femural pulse. Infants and children with aortic coarctation were operated and followed up. In two children we found re-coarctation and high blood pressure. This patients started antihypertensive treatment and followed interventional balloon dilatation. One case was stent implanted, the other no because of a gothic aortic arch.

Conclusions: Absent femural pulse associated with high blood pressure in upper arms can suspect the diagnose of aortic coarctation. Post surgery re-coarctation has to be followed. Interventional balloon dilatation and stent implantation could be necessary in this situation. Even so, patients has to be followed up because high blood pressure can persist and has to be medicated.

2. EVOLUȚIA PACIENȚILOR CU VENTRICUL DREPT SISTEMIC

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Introducere.

Pacienţii cu circulaţie biventriculară la care ventriculul drept este ventriculul sistemic (VDs), pot avea diverse complicaţii şi fiecare pacient beneficiază de soluţii terapeutice specifice.

Obiective.

Urmărirea aspectelor evolutive şi terapeutice la pacienţii cu VDs.

Material şi metodă.

Douăzeci şi unu de copii cu VDs au fost urmăriţi pe o perioadă între 1 şi 6 ani. Subiecţii au fost grupaţi în trei categorii: 1) D-transpoziţie de vase mari (TMV) cu switch arterial, 2) D-TMV cu switch atrial, 3) TMV corectată congenital. S-a efectuat studiu clinic, rezultatele fiind prelucrate şi raportate la datele publicate.

Rezultate.

Dintre cei 21 subiecţi, 19 au prezentat D-TMV cu asocieri malformative cardiace şi 2, TMV corectată congenital. Dintre pacienţii cu D-TMV, 16 au beneficiat de swicth arterial în perioada neonatală şi 3, swicth atrial, după 4 luni de viaţă. Evoluţia subiecţilor operaţi în perioada neonatală a fost marcată de două complicaţii importante în perioada precoce preoperatorie

(hidrocefalie acută, peritonită). Ceilalți au evoluat favorabil. Subiecții cu D-TMV care au primit switch atrial au avut complicaţii: tulburări de ritm supraventriculare şi disfuncţie ventriculară dreaptă. Evoluția clinică a pacienţilor cu TMV corectată congenital a fost marcată de insuficiență cardiacă tardivă. Terapia medicală s-a adresat complicațiilor și s-a efectuat conform protocoalelor AHA.
Concluzii.
Copiii cu VDs de etiologie D-TMV cu switch arterial precoce au evoluție clinică și imagistică favorabilă la distanță. Evoluția subiecților cu VDs prin D-TMV ce au primit corecție prin switch atrial și a celor cu TMV corectată congenital este marcată de complicații.

SYSTEMIC RIGHT VENTRICLE – MONITORING THE EVOLUTION

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Introduction.
Patients with biventricular circulation in whom the right ventricle is the systemic ventricle (SRV) may have various complications and each patient benefits from specific therapeutic solutions.
Objectives.
Monitoring evolution and therapeutic aspects in patients with SRV.
Material and method.
Twenty-one children with SRV were monitored over a period between 1 and 6 years. The subjects were divided into three categories: 1) D-transposition of the great arteries (TGA) with arterial switch, 2) D-TGA with atrial switch, 3) congenitaly corrected TGA. A clinical study was performed and the results were processed and correlated with the published data.
Results.
Of the 21 subjects, 19 had D-TGA associated with cardiac malformations and 2 had congenitally corrected TGA. Of the patients with D-TGA, 16 had arterial switch in the neonatal period and 3 had atrial switch after 4 months of life. The evolution of the subjects operated in the neonatal period was marked by two important complications in the early preoperative period (acute hydrocephalus, peritonitis). The other patients had a favorable evolution. Subjects with D-TGA who had atrial switch developed complications: supraventricular rhythm disorders and right ventricular dysfunction. The clinical evolution of patients with congenitally corrected TGA was marked by late heart failure. Medical therapy was intended for complications and was performed according to AHA protocols.
Conclusions.
Children with SRV of D-TGA etiology with early arterial switch have a favorable long-term clinical and imaging evolution. The evolution of subjects with SRV through D-TGA with atrial switch correction and of those with congenitally corrected TGA is marked by complications.

3. STECUSTICA APROAPE SIMILARA, SITUATII MORFOLOGICE DIAMETRAL OPUSE

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Malformatiile cardiace sunt diagosticate adesea dupa ce au trecut de filtrul neonatal sau in urma unui examen pediatric de rutina. Decelarea unui suflu, completat de investigatii paraclinice si imagistice, permite incadrarea intr-o patologie organica sau functionala.
Experienta clinica, acuratetea examenului obiectiv pot orienta asupra tipului lezional, dar multe variante morfologice pot avea aceeasi stetacustica. De exemplu defectul septal interventricular si stenoza pulmonara- similar stetacustic, dar cu suferinta clinica diferita.
Prezentam cazurile a doi pacienti de 17, respectiv 10 ani, care la examenul obiectiv prezentau un suflu sistolo-diastolic grad IV/VI, stetacustica orientand catre un canal arterial permeabil. Pacienta adolescenta a fost diagnosticata in prima copilarie cu malformatie valvulara, dispensarizata necorespunzator aproximativ 10 ani; a dezvoltat un proces endocarditic la varsta de 15 ani, care a compromis aparatul valvular si a generat insuficienta cardiaca marcata. Al doilea pacient provine din mediu social precar (institutionalizat), cu suspiciune de malformatie cardiaca neelucidata pana la internarea in Clinica.
Echocardiografic s-a decelat substratul morphologic real, incadrarea intr-un tip lezional malformativ diferit si a indicat abordarea terapeutica diametral opusa.
Concluzii:

1. Uneori-in cazul leziunilor rare- stetacustica poate orienta pe cai false.
2. Imagistica in diagnosticul malformatiilor de cord este suverana.
3. Suntem curiosi cum va fi stetacustica acestor pacienti dupa rezolvarea chirurgicala (“Mai seamana?”).

Cuvinte cheie: suflu, malformatie cardiaca, echocardiografie

SIMILAR SOUNDS, OPPOSITE MORPHOLOGICAL SITUATIONS

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Cardiac malformations are often diagnosed by a routine pediatric exam, most of them passing undetected through the neonatal “filter” . The detection of a heart murmur, with the added support of imaging and laboratory investigations, allows the condition to be classified as eider functional or organic.

The clinical experience associated with a well conducted physical examination can guide the physician in respect to the lesion type, but many morphological variants can have the same sounds. For example, interseptal ventricular defect and pulmonary stenosis have similar sounds but are very different clinical conditions.

We are presenting the cases of two patients 17 and 10 years old witch during the physical examination showed systolic-diastolic murmur grade IV / VI, suggesting a permeable arterial channel. The first patient was diagnosed in early infancy with valvular malformation, inappropriately followed for about 10 years; at 15 years of age she developed a endocarditic process witch compromised the hole valvular apparatus and resulted in marked heart failure. The second patient comes from difficult social environment (institutionalized) with suspected cardiac malformation unsolved until hospitalization in our clinic.

Echocardiography was used to establish the real morphologic substrate, resulted in the classification of the malformation type quite different from the first case and indicated a diametrically oppose therapeutic approach.

Conclusions:

1. Sometimes, in the case of rare lesions- the sounds can suggest false pathology.
2. The value of imaging in the diagnosis of heart malformations is sovereign.
3. We are curious how it will these patients sound after surgical resolution ("still resemble?").

Key words: murmur, cardiac malformation, echocardiography

ENDOCRINOLOGIE ŞI DIABET PEDIATRIC / PEDIATRIC ENDOCRINOLOGY AND DIABETES

4. BARIERE IN REALIZAREA ECHILIBRUL GLICEMIC LA COPIII ŞI ADOLESCENŢII CU DIABET ZAHARAT TIP 1

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Introducere. Desi in ultimii ani s-au folosit tehnologii noi si tipuri noi de insuline, studiile nu au evidentiat o imbunatatire semnificativa a echilibrului glicemic in diabetul zaharat (DZ) tip 1, ceea ce sugereaza ca pe langa metodele mai noi de tratament si monitorizare, exista si alti factori care influenteaza echilibrul glicemic.

Obiectiv. In acest studiu am urmarit stabilirea unor corelatii intre echilibrul glicemic si anumiti parametri la copiii si adolescentii cu DZ tip 1.

Material şi metoda. Studiul s-a efectuat asupra unui lot de 41 copii si adolescenti cu DZ tip 1, pe baza unui chestionar care a fost completat de catre parinti. Echilibrul glicemic a fost definit la valori ale hemoglobinei glicate (HbA1c) sub 7,5%.

Rezultate. Principalii factori care s-au corelat cu un echilibru glicemic scazut la copiii si adolescentii cu DZ tip 1 luati in studiu au fost: grupa de varsta pubertara (72,73%), pacientii proveniti din mediul rural (68,43%), existenta unor conflicte familiale permanente (100%), pacientii care efectueaza exercitii fizice sporadic (92,86%), pacientii care nu respecta cantarirea alimentelor la mese (80%), copiii a caror parinţi au doar studii gimnaziale (76,46%), copiii aflati in primul an dupa debutul diabetului (60%), copiii la care s-au implicat doar unul din parinti in ingrijirea diabetului (61,54%), frecventa determinarilor glicemiei sub 4/zi (81,82%). S-au mai stabilit corelatii pozitive intre valorile taliei, greutatii, IMC si valorile HbA1c.

Concluzii. La majoritatea copiilor, un controlul glicemic bun poate fi realizat printr-o mai buna intelegere a bolii si a factorilor care influentează echilibrul glicemic.

Cuvinte cheie: echilibru glicemic, copil si adolescent, diabet.

BARRIERS IN GLYCEMIC CONTROL IN CHILDREN AND ADOLESCENTS WITH TYPE 1 DIABETES

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Introduction. Although in recent years new technologies and new types of insulin have been used, no significant improvement has been observed in glycemic control of type 1 diabetes (T1D). This suggests that apart from the new methods of treatment and monitoring, there are also other factors that influence glycemic control.
Aim. In this research we have studied the existence of correlations between glycemic control and certain parameters in children and adolescents with T1D.
Material and method. The study was performed on a group of 41 children and adolescents with T1D, based on a questionnaire which was filled in by the parents. Optimal glycemic control was reached when the value of glycated hemoglobin (HbA1c) was below 7.5%.
Results. The main factors that were correlated with the deterioration of glycemic control in the children and adolescents with T1D who participated in the study were: pubertal age groups (72.73%), patients from rural areas (68.43%), the existence of permanent family conflicts (100%), patients who rarely exercise (92.86%), patients who do not weigh food (80%), patients whose parents have only secondary education (76.46%), patients in the first year after onset (60%), patients that had only one parent involved in diabetes care (61.54%), patients with less then 4 glycemic measurements per day (81.82%). Positive correlations have also been established between height, weight, body mass index and HbA1c.
Conslusions. At most children, an optimal glycemic control can be achieved through a better understanding of the disease and the factors influencing glycemic control.
Key words: glycemic control, child and adolescent, diabetes

5. DEZORDINILE DE DEZVOLTARE SEXUALA - SOLUTII MICI LA PROBLEM MARI

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Introducere: Dezordini de Dezvoltare Sexuala (DSD) reprezinta o realitate a practicii medicale regasite cu o prevalenta de 1 la 300 de copii
Material si metoda: In perioada 2010-2014 am studiat un numar de 31 pacienti cu DSD. Investigatiile complexe au vizat anamneza, examen clinic obiectiv;biologic s-au efectuat explorari complexe (ionograma sanguina, 17- OHprogesteron, LH, FSH, estrogeni, testosteron, DHEA,DHEA-s, cariotip,la care s-au adaugat explorari suplimentare in functie de caz; explorari imagistice (in functie de forma).
Rezultate si discutii: Pe o perioada mentionata am diagnosticat 31 pacienti cu DSD, respectiv 6 cu deficit de 21hidroxilaza forma cu pierdere de sare ;1 pacient cu deficit de 17 alfa hidroxilaza si 4 pacienti cu 46XY DSD. 20 de pacienti au prezentat 46 XX DSD. 2 cazuri au fost diagnosticate prin screening neonatal derulat prin proiectul European Hu-Ro 0802/008 SCREENGEN. Cariotipul a fost definitor in creionarea sex-ului genetic. Pentru adolescentii 46XY DSD diagnosticul realizat, a fost extrem de greu de explicat familiei si pacientilor. Tratamentul a vizat substitutia cu hidrocortizon pentru pacientii cu deficit de 21hidroxilaza si substitutie estro-progestaiva pt pacientii 46 XY DSD. Remodelarea chirurgicala s-a realizat in cazul celor cu clitoromegalie si/sau 46XY DSD. Suportul psihologic a fost necesar in toate cazurile. Unele familii au contestat diagnosticul acesta fiind confirmat de clinici din Europa. Abordarea anomaliilor se realizaeaza in echipa.
Concluzii:

1. Prevalenta anomaliilor de dezvoltare sexuala este mare si subdiagnosticata
2. Este necesara o echipa complexa interdisciplinara pentru managementul correct al anomaliei
3. Este imperios necesara realizarea registrului national DSD .

SEXUAL DEVELOPMENT DISORDER - SMALL SOLUTIONS TO BIG PROBLEM

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Results and discussion: Over a period mentioned we diagnosed 31 patients with DSD. 6 with 21hidroxilaza deficit with loss of salt form, 1 patient with 17 alpha hydroxylase deficiency and 4 patients with 46XY DSD. 20 patients experienced 46 XX DSD. 2 cases were diagnosed through neonatal screening conducted by the European project SCREENGEN Hu-Ro 0802/008. Karyotype was definitively outlining's genetic sex. For teenagers 46XY DSD diagnosis was made but extremely difficult was to explain to family and patients this situation. Hydrocortisone replacement therapy

targeted to patients with impaired 21hidroxilaza and substitute estro-progestaivs for 46 XY DSD patients. Surgical remodeling was done in those with clitoromegalie and / or 46XY DSD. Psychological support was necessary in all cases. Some families have challenged this diagnosis was confirmed by clinics in Europe. Addressing anomalies was done in teams.
Conclusions:
1. The prevalence of sexual development abnormalities is high and underdiagnosed
2. It requires complex interdisciplinary team to correct the anomaly management
3. It is necessary to create a national registry DSD.

6. RELATIA DINTRE INDICELE DE MASA CORPORALA, PROCENTUL DE TESUT ADIPOS SI NIVELUL LEPTINEI LA COPIII OBEZI

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Obezitatea este o tulburare a starii de nutritie, caracterizata prin acumularea excesiva de grasimi in tesutul celular subcutanat si/sau alte tesuturi ca rezultat al dezechilibrului balantei energetice. Leptina , principalul hormon anorexigen, este secretat de tesutul adipos si actioneaza asupra centrilor satietatii si foamei din hipotalamusul anterior si nucleul lateral. Obezitatea se caracterizeaza cu ajutorul indicelui de masa corporala (IMC) raportat la sex si varsta.
Obiectivul acestui studiu a fost reprezentat de analiza corelatiei dintre indicele de masa corporala, procentul/cantitatea de tesut adipos si nivelul seric al leptinei la copiii cu exces ponderal.
Materialul a fost reprezentat de 126 copii (86 cu exces ponderal - lotul de studiu, si 40 cu greutate normal - lotul martor) cu varsta intre 4 -18 ani, internati in ultimii 2 ani (in perioada 2013-2015) în Compartimentul de Genetica Medicala al Spitalului Clinic de Urgenta pentru Copii din Cluj-Napoca.
Metoda de lucru a inclus: anamnaza; examen clinic; măsurători antropometrice (greutate, talie, IMC); determinarea procentul de tesut adipos prin metoda analizei impedantei bioelectrice (Omron); teste biochimice: profil lipidic, glicemie; dozari hormonale: insulinemie, cortisol si leptina.
Rezultate: Obezitatea predomina la sexul feminin (58,6%) fata de cel masculin (41,4%) in lotul studiat . Analiza compozitiei corporale a aratat un procent mai mare al tesutului adipos la sexul feminin. Nivelul leptinei se coreleaza pozitiv cu IMC si procentul de tesut adipos, sex si varsta.
Concluzii: Nivelul crescut al leptinei la copiii cu obezitate contribuie la dezvoltarea precoce al sindromului metabolic.
Cuvinte cheie: obezitate, IMC, procent de tesut adipos, leptina.

RELATIONSHIP BETWEEN THE BODY MASS INDEX, PERCENTAGE OF ADIPOSE TISSUE AND LEPTIN IN OBESE CHILDREN

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Obesity is a disorder of nutritional status, characterized by excessive accumulation of fat in the subcutaneous cellular tissue and / or other tissue as a result of energy imbalance. Leptin is the main anorexigenic/catabolic hormone, it reduces food intake and increases the activity of the sympathetic nervous system. It is secreted by adipose tissue and acts on ventromedial hypothalamus and lateral hypothalamic area. Obesity is characterized using the body mass index (BMI) according to gender and age.

The aim of this study was to analyze the correlation between body mass index, percentage/amount of adipose tissue and leptin serum levels in overweight children.

The material was represented by 126 children (86 with excess weight - the study group, and 40 normal weight - control group) aged 4 -18 years, hospitalized in the last 2 years (2013-2015) in the Department of Genetics Medical Emergency Hospital for Children in Cluj-Napoca.

The working method included: anamnesis; clinical examination; anthropometric measurements (Weight, Height, BMI); determining the percentage of body fat by bioelectrical impedance analysis method (Omron); biochemical tests: lipid profile, glucose; hormonal assays: leptin, insulinemie and cortisol.

Results: Obesity is more common in girls (58.6%) than boys (41.4%) in the study group. The analysis of the body composition showed a higher percentage of body fat in females. The leptin level is positively correlated to BMI, percentage of body fat, sex and age.
Conclusions: Increased serum levels of leptin in obese children contributes to early development of metabolic syndrome.
Keywords: obesity, BMI, body fat percentage, leptin

GENETICĂ / GENETICS

7. CORELAȚII GENOTIP-FENOTIP LA PACIENȚII CU SINDROM BECKWITH-WIEDEMANN

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Sindromul Beckwith-Wiedemann (BWS), o boala genetică rara este cauzata de mutatia sau deletia genelor amprenate de la nivelul regiunii cromozomiale 11p15.5. BWS se caracterizeaza prin hemihipertrofie, macroglosie, defecte la nivelul peretelui abdominal, hipoglicemie neonatala, predispozitie pentru aparitia tumorilor.
Am investigat corelatiile genotip-fenotip la 5 pacienti cu doagnostic clinic de sindrom BS care au fost referiti Cabinetului de genetica din cadrul Spitalului Judetean de Urgeta Tirgu Mures intre 2014-2015. La toate cazurile s-a efectuat analiza citogenetica, aceasta neevidentinand modificari cromozomiale in timp ce analiza de metilare (MS-MLPA) a aratat hipometilarea la nivelul centrului de amprentare IC. Pentru analizele genetice s-a folosit sange proaspat de la pacientii cu BWS. La pacientii nostrii cu BWS nivelul alfa-fetoproteinei (AFP) a fost mai mare decat valorile normale. Am observat o asociere intre macroglosie, nivel de AFP crescut si hipometilarea IC2 (KvDMR).
Cuvinte cheie: Beckwith-Wiedemann, analiza de metilare, corelatii

GENOTYPE–PHENOTYPE CORRELATIONS IN PATIENTS WITH BECKWITH–WIEDEMANN SYNDROME

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Beckwith-Wiedemann syndrome (BWS), a rare genetic disorder is caused by mutation or deletion of imprinted genes within the chromosome 11p15.5 region. BWS is caractherised by hemihypertrophy, macroglossia, abdominal wall defects, visceromegaly, hypoglycemia neonatally, predisposition to tumor development.
We investigated genotype-phenotype correlations in 5 patients with clinical diagnostic of BWS who were reffered to Genetics Depatment of Emergency County Hospital Tirgu Mures between 2014-2015. Cytogenetic analyses were performed in all cases and revealed no chromosomal abnormalities while methylation anayles (MS-MLPA) showed ICR hypomethylation. Genetic analyses in BWS were performed by using fresh peripheral blood. In all BWS patients alpha-fetoprotein (AFP) concentration was anaylsed. AFP levels obtained inour patients were higher than normal value. We observed an association between macroglossia, high levels of AFP and IC2 hypomethylation (KvDMR).
Keywords: Beckwith-Wiedemann, methylation anayles, correlations

8. INCIDENTA PRINCIPALELOR MUTATII ALE GENELOR GJB2 SI GJB6 IN SURDITATEA NEUROSENZORIALA DIN ROMANIA.

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Obiectiv. Studiul de fata si-a propus sa analizeze (pe un lot reprezentativ statistic) incidenta celor mai frecvente mutatii ale genelor GJB2 si GJB6 la pacientii cu surditate congenitala non-sindromica.
Metoda : 179 copii neinruditi cu surditate congenitala non-sindromica au fost evaluati prin urmatoarele metode: a. Examen audiologic; b. Detectarea mutatiilor c.35delG si pW24X (prin tehnica semi-nested PCR urmata de analiza RFLP si ARMS-PCR); c.Detectarea mutatiilor del(GJB6-D13S1830) si del (GJB6-D13S1854) prin tehnica PCR-multiplex.
Rezultate si discutii: Analiza celor 4 mutatii ale genelor GJB2 si GJB6 a permis stabilirea cauzei genetice a surditatii la 45 pacienti (25.14% din totalul lotului). Incidenta mutatiilor a fost de 27.3 % pentru c.35delG, 3.6 % pentru p.W24X și 0.28% pentru del(GJB6-D13S1830). Nu a fost depistat nici un caz cu mutatia del (GJB6-D13S1854).
Concluzii: Incidentele celor patru mutatii analizate la pacientii romani cu surditate congenitala non-sindromica sunt comparabile cu cele depistate in alte tari Europene cu populatie non-latina. Pentru o interventie terapeutica precoce este obligatorie efectuarea (la nou-nascuti) a screeningului genetic universal sau a screeningului audiologic urmat de testarea genetica la cazurile cu hipoacuzie.

INCIDENCE OF MUTATIONS OF GJB2 AND GJB6 GENES IN NON-SYNDROMIC HEARING LOSS OF ROMANIA.

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Objective. To determine the frequency of occurrence of the most common mutations of *GJB2* and *GJB6* genes in patients with congenital nonsyndromic sensorineural hearing loss (NSHL).
Methods : 179 unrelated Romanian children with congenital NSHL have been evaluated with: a. audiological examination; b.Detection of *c.35delG* and *pW24X* mutations (semi-nested PCR technique followed by RFLP as well as ARMS-PCR analysis); c.Detection of *del(GJB6-D13S1830)* and *del (GJB6-D13S1854)* by PCR-multiplex technique.
Results: Audiological examination determined a diagnosis of profound, severe or moderate hearing loss in 116, 29 and 34 patients respectively, representing 64.8%, 16.2% and 19% of the trial patients, respectively. The analysis of the 4 mutations of genes *GJB2* and *GJB6* has allowed the determination of the genetic cause for hearing loss in 45 patients (25.14% of the lot). The frequency of occurrence was 27.3 % for *c.35delG*, 3.6 % for *p.W24X* and 0.28% for *del(GJB6-D13S1830).* No case with *del (GJB6-D13S1854)* was found in the ample group.
Conclusions: The frequency rate of the four mutations analyzed in patients with NSHL from Romania is comparable to that found in other studies. For an early therapeutic intervention, in the case of newborns, the universal genetic screening or the audiological screening followed by genetic testing in confirmed congenital hypoacusis cases, are compulsory.

HEMATOLOGIE ŞI ONCOLOGIE / HAEMATOLOGY AND ONCOLOGY

9. TRATAMENTUL MULTIDISCIPLINAR MODERN IN TUMORILE CU CELULE GERMINALE INTRACRANIENE

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Introducere. Tumorile germinale intracraniene (TGC) apar predilect la copii si adolescenti (70% intre 10- 21 ani). Interesul deosebit pentru aceste tumori rezulta din faptul ca se pot vindeca in procente de 75- 95% prin tratament multidisciplinar modern. Chimioterapia este parte integrala a tratamentului tumorilor germinale cu alta localizare decat sistemul nervos central. Ratiunea utilizarii chimioterapiei in tratamentul TGC este de a reduce efectele secundare la distanta ale iradierii cerebrale prin scaderea dozei si campului de iradiere in tumorile localizate si de a imbunatati calitatea supravietuirii in cazurile metastazate.
Material si metode. Sunt prezentate 22 de cazuri tratate in Institutul Oncologic Bucuresti in intervalul 2004- 2012. Diagnosticul de certitudine s-a stabilit prin biopsie sau cresterea markerilor specifici (alfa 1 fetoproteina, hormon gonadotrop corionic). Protocolul de tratament utilizat a fost protocolul propus de Societatea Internationala de Oncologie Pediatrica : chirurgie limitata (doar biopsie) urmata de Chimioterapie (4 serii) si Radioterapie (camp cranian 24- 30 Gy in cazul rspunsului complet sau 30- 36 Gy pentru raspuns partial, iradiere spinala in cazurile in care exista celule maligne in LCR).
Rezultate. Toti pacientii au fost evaluati clinic, biologic si imagistic dupa 2 si/sau 4 serii de chimioterapie si post-radioterapie. Urmarirea clinica si paraclinica s-a facut cel putin 2 ani de la finalizarea tratamentului. Rata estimata de supravietuire globala la 5 ani a fost de 82%.
Concluzii: 1. Chimioterapia policitostatica este eficienta in tumorile germinale intracraniene, atat in scop curativ cat si paliativ.2. Rezultate terapeutice excelente in cazurile tratate cu chimio si radioterapie dupa interventie chirurgicala limitata (diagnostica).
Cuvinte cheie: tumori germinale, copil, tratament modern

THE MODERN MULTIDISCIPLINARY TREATMENT IN INTRACRANIAL GERM CELL TUMORS

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Introduction. Intracranial germ cell tumors (TGC) arrise especially in children, adolescents and (70% between 10-21 year old). These group of tumors request special interest because 75-95% of them can be cured with a modern multimodal treatment. Chemotherapy is an integral part of the multimodal treatment of the germ cell tumors located outside the central nervous system. The reason for using chemotherapy for intracranial germ cell tumors is to reduce the total dose and the target field for radiotherapy in order to reduce the incidence of late side effects and to improve the quality of life for patients with metastasis.
Material and methods. There are presented 16 patients treated in the Institute of Oncology Bucharest between 2004-2012. The diagnosis was established by biopsy or elevated specific tumor markers (alpha1-fetoproteine, human-chorionic-

gonadotropin). The treatment (according to the protocol proposed by the International Society of Pediatric Oncology) consists in limited surgery (only biopsy), followed by Chemotherapy (4 cycles) and Radiotherapy (cranian field 24-30Gy after complete response or 30-36G after partial response and spinal irradiation in cases with malignant cell in the CSF).
Results: All of the patients were assessed clinical, biological and imagistical after 2 and/or 4 cycles of chemotherapy and after radiotherapy . Clinical and paraclinical follow-up at least 2 years after the completion of treatment . The estimated overall survival rate at 5 years 82%.
Conclusions: 1. Chemotherapy is efficient in intracranial germ cell tumors either in curative or palliative purpose. 2. Excellent therapeutic results with chemotherapy and radiotherapy after only diagnostic surgery.
Keywords: germ cell tumors, children, modern treatment

10. PROFILUL COMPLICATIILOR INFECTIOASE LA PACIENTII IMUNODEPRIMATI

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Introducere:Scopul acestui studiu il reprezinta analiza profilului complicatiilor infectioase la o categorie de pacienti cu deficit imun sever – populatia pediatrica cu transplant de celule stem hematopoietice (TCSH) autolog si/sau allogeneic.
Pacienti si metode:Intr-un studiu retrospective am inclus toti copiii transplantati autolog (n=82) sau allogeneic (n=27) in Centrul de Transplant Medular Timisoara. Pe langa masurile standard privind profilaxia si diagnosticul infectiilor, au fost monitorizate probele inflamatorii (CRP, PCT si IL-6) precum si reactivarea moleculara a virusurilor *Herpesviridae* (la pacientii cu TCSH allogeneic).
Rezultate:Culturile cutaneo-mucoase recoltate pretransplant au fost pozitive la 39.44% din pacienti: 81.39% cu germeni gram-pozitivi, 18.6% cu germeni gram-negativi iar 3.66% cu fungi. In total, s-au inregistrat 122 de episoade de neutropenie febrila (NF), 14.75% fiind documentate microbiologic iar 9.01% doar clinic. Spre deosebire de colonizarea cutaneo-mucoasa, germenii izolati in cadrul episoadelor de NF au fost predominant gram-negativi (88.9%). Infectiile fungice probabile sau posibile au fost diagnosticate la 3.66% din pacienti. Valorile maxime ale CRP si PCT au fost predictive pentru etiologia bacteriana a episoadelor de NF, cu un coeficient inalt de corelatie (0.64). La 33.33 % din pacientii cu TCSH allogeneic, a fost depistat molecular cel putin un episod de reactivare a virusurilor din familia *Herpesviridae,* PCR pozitivandu-se cel mai frecvent pentru CMV (25.92%).
Concluzie:Complicatiile infectioase influenteaza negative prognosticul HSCT, imbunatatirea sperantei de viata dupa TCSH fiind posibila prin optimizarea masurilorprofilactice, a diagnosticului precoce precum si printr-o terapie antimicrobiana tintita.

THE PROFILE OF INFECTIOUS COMPLICATIONS IN IMMUNOCOMPROMISED PEDIATRIC PATIENTS

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Introduction:We aimed to analyze the profile of infections in a severely immunosuppressed population–children with autologous and/or allogeneic hematopoietic stem cell transplantation (HSCT).
Patients and methods:In a retrospective study we have included all the pediatric patients transplanted autologous (n=82) or allogeneic (n=27) in the BMT Center Timisoara. Standard measures for prevention and diagnosis of infectious were adopted. In addition, monitoring of inflamatory cytokines, as well as molecular monitoring for *Herpesviridae* (in allogeneic HSCT) were performed.
Results:Pretransplant swabs screening revealed a mucosal and/or cutaneous colonization in 39.44% of the patients: 81.39% with gram-positive, 18.6% with gram-negative strains and 3.66% with fungi. Overall, 122 febrile neutropenic (FN) episodes were registered, 14.75% of them being microbiologically-documented whereas 9.01% were clinically-documented infections. In discordance with the colonization, the profile of agents isolated in FN episodes was dominated by gram-negative strains (88.9%). Probable and/or possible invasive fungal infections were noted in 3.66% of the patients. Maximum CRP and maximum PCT levels registered during the FN episodes were predictive for the bacterial etiology of FN whit a high correlation (0.64). At least one episode of active infection due to *Herpesviridae* occurred in 33.33% of the patients with allogeneic HSCT, CMV being the most frequent viral pathogen detected (25.92%).
Conclusion:Infectious complications negatively influence the outcome of HSCT, preventive measures and early identification as well as a specifically tailored therapy being only some of the requirments in the attemp to increase the life expectancy after HSCT.

11. HEMOLIMFOHISTIOCITOZA – O AFECTIUNE RARA CU PROGNOSTIC REZERVAT

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Introducere: Hemolimfohistiocitoza (HLH) este un sindrom care asociaza semne clinice si anomalii de laborator secundare proliferarii si activarii necontrolate a celulelor liniei monocito- macrofagice. HLH se prezinta fie ca forma

familiala (FHL) fie secundara (sHLH) asociata unor boli infectioase (in special infectia EBV), maligne sau reumatismale. Cele 2 forme nu au
manifestari clinice distincte. FHL este rapid fatala in lipsa tratamentului specific.
Scop: analiza rezultatelor tratamentului HLH.
Pacienti si metoda: criteriile revizuite 2004 au fost utilizate pentru diagnostic; toti pacientii au efectuat protocolul HLH 2004. Doi pacienti au efectuat transplant cu conditionare non-
mieloablativa cu donator neinrudit.
Rezultate: in Institutul clinic Fundeni, in perioada 2000-2014 au fost diagnosticati 9 pacienti cu forme severe de HLH. Patru cazuri de FHL (Munch 13 (1), XIAP2(1), anomalii netestate (2)), 5 cazuri de sHLH (induse EBV (4), asociate malignitatii (1)). 2/4 FHL au efectuat transplant si sunt in viata, cu afectare neurologica. 4/5 cazuri de sHLH au raspuns initial la terapia specifica. Am inregistrat 2 decese in grupul FHL si 3 in grupul sHLH.
Discutii/Concluzii: FHL este diagnosticat in primele luni de viata, triggerul find o infectie; FHL asociat cu sindromul limfoproliferativ X linkat poate fi diagnosticat la baieti pana in perioada adolescentei; sHLH poate fi diagnosticat la orice varsta. Determinarea valorilor feritinei si a trigliceridelor este obligatorie pentru suspiciunea de HLH. Transplantul medular reprezinta singura terapie curativa pentru FHL si sHLH la recadere. Diagnosticul rapid al HLH este foarte
important pentru prognostic.

HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS – A RARE DISEASE WITH POTENTIAL UNFAVORABLE OUTCOME

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Background: Hemophagocytic lymphohistiocytosis (HLH) is a syndrome consisting of clinical signs and laboratory abnormalities that result from uncontrolled proliferation and activation of cells of the monocyte/macrophage lineage. HLH is categorized in 2 forms- familial (FHL) and secondary (sHLH)- indistinguishable by clinical presentation. SHLH is associated with infection, malignancy, rheumatologic diseases. FHL is rapidly fatal without specific therapy.
Aim: to analyze the results of therapy for HLH.
Patients and methods: the revised guidelines 2004 were used for diagnostic; all patients received treatment according to HLH-2004 protocol. Two patients received stem cell transplantation (SCT) reduced intensity from unrelated donors.
Results: In Fundeni Clinical Institute we diagnosed 9 cases of severe HLH between 2000-2014. There were 4 cases of FHL (Munc 13 (1), XIAP2 (1), unknown abnormalities (2)) and 5 cases of sHLH (EBV induced (4), malignancy associated (1)). All patients received HLH-2004 protocol; 2/4 FHL were transplanted and alive, with neurological impairment. In the sHLH group, 4/5 cases initially responded after specific therapy. We registered 2 deaths in the FHL group due to disease progression and 3 deaths in the sHLH group due to malignancy evolution (1), HLH progression (2).
Discussion/Conclusion: FHL is diagnosed in the first months and it's triggered by an infection; the FHL associated with X lymphoproliferative syndrome could be diagnosed in late childhood or adolescence in boys. sHLH could be diagnosed at any time. SCT is the only curative therapy for FHL or for relapsed sHLH. The rapid diagnosis of HLH is very important for outcome.

IMUNOLOGIE / IMMUNOLOGY

12. RELATIA ANEMIE FERIPRIVA - IMUNITATE UMORALA

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Introducere: Anemia feripriva este cea mai frecventa boala hematologica a copilului mic. Rolul fierului in reglarea imuna nu este inca bine cunoscut. Obiectivul studiului: evaluarea efectelor anemiei feriprive si a tratamentului cu fier asupra imunitatii umorale.
Material si metoda: In lotul de studiu au fost inclusi 100 copii, cu varsta 1-3 ani diagnosticati cu anemie feripriva in Clinica III Pediatrie - Spitalul Clinic de Urgenta pentru copii "Sfanta Maria" Iasi. Criteriile de includere au fost: hemoglobina (Hb) < 11 g/dl, VEM< 70μ^3, HEM < 25 pg, feritina< 20 ng/ml, fierul sub valorile normale. Criterii de excludere: copii care au primit tratament cu fier sau multivitamine in ultimele 3 luni, prezenta infectiilor acute, malnutritiei, bolilor cronice. Tratament

administrat: 6 mg/kg/zi fier elemental, 3 luni. Lotul martor :100 copii de aceeasi varsta cu alte afectiuni, dar fara anemie, infectii acute, boli cronice sau malnutritie. La ambele loturi au fost determinate Ig G,M si A.
Rezultate: 12% ditre cazuri au prezentat anemie severa (Hb<7g/dl), 63% -medie (Hb=7-10 g/dl), 25%- usoara (Hb=10-11 g/dl). Valoarea medie a IgM=35 mg/dl la lotul de studiu a fost usor mai scazuta comparativ cu lotul martor,Ig M=50mg/dl . Valori mai scazute ale parametrilor hematologici s-au corelat cu valori mai scazute ale IgG. A fost gasita o corelatie si intre IgA si IgG.
Concluzie: intelegerea relatiei dintre deficitul de fier si alterarea sistemului imun este foarte importanta.
Cuvinte cheie**:** anemie, fier, imunoglobuline, tratament.

RELATIONSHIP BETWEEN IRON DEFICIENCY ANEMIA AND HUMORAL IMMUNITY

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Introduction: Iron deficiency anemia is the most common hematologic disease at small children. The role of iron in immune regulation is still not well known. The objective of the study: assessment the effects of the treatment of iron deficiency anemia and iron on humoral immunity .
Materials and methods: The study group included 100 children, aged 1-3 years diagnosed with iron deficiency anemia in Pediatrics Clinic III - Emergency Hospital for Children " Saint Mary" Iasi. Inclusion criteria were: hemoglobin (Hb) < 11 g / dl, MCV < 70µ3 , MCH < 25 pg , ferritin < 20 ng / ml , iron below normal. Exclusion criteria : children who received iron or multivitamins in the last 3 months , acute infections , malnutrition , chronic disease . Treatment administered 6 mg / kg / day of elemental iron , 3 months. Control group : 100 children the same age , but without anemia. Both groups were determined Ig G, M and A.
Results : 12% cases had severe anemia (Hb < 7g / dl), 63%-moderate anemia (Hb = 10.07 g / dl), 25 % - mild anemia(Hb = 11.10 g / dl) .Mean IgM = 35 mg / dL was decreased compared with controls , mean Ig M = 50mg / dl. Lower values of hematological parameters were correlated with lower levels of IgG .
Conclusion: understanding the relationship between iron deficiency and impaired immune system is very important
Keywords: anemia , iron, immunoglobulin, treatment.

13. DEFICITUL SELECTIV PRIMAR DE IgA SIMPTOMATIC LA COPIL

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Deficitul primar selectiv de IgA este o imunodeficienţă ce apare la copiii mai mari de 4 ani, care prezintă nivele serice scăzute de IgA, sub 7 mg/dl. Cele mai multe cazuri sunt asimptomatice însă sunt citate asocieri cu infecţiile respiratorii recurente, alergiile, bolile autoimune şi neoplaziile.
Prezentăm cazul unui pacient de 7 ani, care s-a adresat Clinicii II Pediatrie a Spitalului Sf.Maria Iaşi, pentru exacerbarea unei tuse matinale, cronice, cu debut insidios, în urma cu un an, cu expectoraţie mucopurulentă şi apariţia a 3 formaţiuni tumorale scapular drept.Copilul prezintă în antecedente numeroase infecţii respiratorii superioare, predominant otite şi inferioare, multe dintre ele cu germeni agresivi, ce au necesitat multiple scheme terapeutice. Diagnosticul a fost confirmat pe baza imunogramei care a aratat valori scăzute ale IgA, cu valori normale ale celorlalte Ig. Elementele clinico-anamnestice coroborate cu datele de laborator, imaginile de bronşiectazii multiple surprinse pe CT toracic si audiograma care descrie o hipoacuzie de transmisie, conturează asocierea unui deficit primar de IgA cu bronşiectazia şi hipoacuzia, amprente ale infecţiilor respiratorii multiple.Tratamentul de substitutie cu Ig iv nu se recomandă, întrucît se pot dezvolta anticorpi împotriva IgA, cu riscul de şoc anafilactic. Prognosticul este grevat de recurenţa şi severitatea episoadelor infecţioase, cu agravarea bronşiectaziei şi fibrozei pulmonare precum şi de confirmarea sau infirmarea caracterului neoplazic tumoral.
Cuvinte cheie: deficit de IgA, infectii respiratorii recurente, bronşienctazie.

SELECTIVE IgA DEFICIENCY SIMPTOMATIC IN CHILDREN

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Selective IgA deficiency is a primary immunodeficiency disease characterized by a decrease of the IgA serum, under 7 mg/dl, in a patient over the age of 4. Most of the IgA deficiency patients are asymptomatic, but some can develop recurrent respiratory infection, allergies, autoimmune condition or cancer.

We give as an example the case of an 7 years old, male child, hospitalized at the 2nd Pediatric Clinic of "St. Maria" Emergency Hospital for children over a year ago, with an exacerbation of a morning chronic cough, with an insidious onset, along with mucopurulent expectoration and a 3 scapular tumors.The child presents a history of a numerous upper respiratory infections, recurrent otitis and lower respiratory infections, many of them with aggressive germs, which require multiple antibiotics.The diagnosis was confirmed based on the immunogram that showed low levels of IgA and normal levels for other immunoglobulin isotypes.
Clinical and anamnesis elements in conjunction with laboratory data, multiple bronchiectasis detected on the thoracic CT and the audiogram which describes transmission hearing loss, emphasizes the association between IgA deficiency with bronchiectasis and the hearing loss, consequences of multiple respiratory infections.
Therapy of replacement with Ig iv is not recommended, because they can develop antibodies against IgA, with risk of anaphylactic shock. Prognosis is encumbered by the recurrence and severity of infectious episodes, with the worsening of lung fibrosis and bronchiectasis, as well as the confirmation or disproval of the neoplazic tumor character.
Keywords: IgA deficiency, recurrent respiratory infections, bronchiectasis.

14. RASPUNSUL BIOLOGIC LA VACCINAREA PNEUMOCOCICA A COPIILOR CU DEFICIT SELECTIV DE IMUNOGLOBULINA A

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Obiective: studiul reactivitatii imune la administrarea unui vaccin cu antigeni polizaharidici, la pacienti cu deficit selectiv de IgA care inregistreaza valori initiale scazute ale anticorpilor specifici antipneumococici.
Material si metode: administrarea unui vaccin pneumococic polivalent neconjugat si urmarirea titrurilor postvaccinale ale anticorpilor secifici antipneumococici, la copii cu deficit selectiv de IgA cu titruri de anticorpi scazute antevaccinal. Pacientii au fost diagnosticati conform criteriilor ESID la Centrul din Debrecen.
Rezultate: la 20 din cei 26 de pacienti vaccinati, tirtrul postvaccinal de anticorpi antipneumococici a crescut la valori normale. La 3 pacienti titrul anticorpilor specifici a ramas nemodificat, iar la alti 3, nivelul anticorpilor, dupa o crestere initiala, a scazut din nou sub limita fiziologica.
Concluzii: majoritatea copiilor cu deficit selectiv de IgA dezvolta o imunitate specifica solida si durabila dupa vaccinarea cu vaccin pneumococic polivalent neconjugat. Totusi, 23% a pacientilor din lotul nostru raman insuficient protejati fata de infectiile pneumococice. Acesti pacienti ar necesita urmarire clinica si biologica individualizata, existand posibilitatea ca diagnosticul sa fie revizuit.
Cuvinte cheie: test vaccinare pneumococica, deficit IgA

BIOLOGICAL REACTIVITY TO PNEUMOCOCCAL TEST VACCINATION IN CHILDREN WITH SELECTIVE IMMUNOGLOBULINE A DEFICIENCY

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Goals: to test the immune reactivity to polysaccharide antigens in patients with symptomatic selective IgA deficiency, with low initial antipneumococcal IgG antibody levels.
Study design: test vaccination of the selected patients with unconjugated polyvalent pneumococcal vaccine and follow-up of the postvacinal specific antibody levels. The patients were diagnosed in the Debrecen Centre.
Results: in 20 of 26 vaccinated patients, the postvaccinal specific antibody levels increased to normal range. In 3 patients we could not find out any incerase, and in other 3 patients, after an initial increase, the antibody levels decreased again below the normal range.
Conclusions: the majority of the sIgAD patients with recurrent respiratory infections are able to develop a strong and durable immunity following a test vaccination with unconjugated polyvalent pneumococcal vaccine. Nevertheless some patients (23%, in our study lot) remain unsufficiently protected when facing pneumococcal infections. These patients need individualized follow-up and their diagnosis may be reconsidered.
Key words: pneumococcal test vaccination, selective IgA deficiency

15. TRANSPLANTUL DE CELULE STEM HEMATOPOIETICE IN IMUNODEFICIENTELE PRIMARE. EXPERIENTA CENTRULUI DE TRANSPLANT TIMISOARA

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In ultimii ani s-au facut pasi importanti in managementul diagnostic si terapeutic al imunodeficientelor primare (IDP) iar utilizarea tehnologiei de secventializare permite o mai buna caracterizare a acestora. Transplantul de celule stem hematopoietice alogenic (TCSH) reprezinta o indicatie consacrata pentru multe IDP letale. Disponibilitatea crescanda a donorilor voluntari de celule stem precum si tehnicile din ce in ce mai rafinate de tipizare HLA au condus la rate de supravietuire de 70-80%. Prezentam experienta Centrului de Transplant din Timisoara in ceea ce priveste TCSH la sase pacienti pediatrici cu IDP. Trei pacienti cu boala granulomatoasa cronica x-linkata (BGC), un pacient cu sindrom Griscelli tip 2 si limfohistiocitoza hemofagocitica (LHH) precum si alti doi gemeni cu imunodeficienta severa combinata (SCID), IL-7 R deficienta, T^- B^+ NK^+ au fost transplantati de la donori inruditi sau neinruditi HLA-compatibili la Centrul de Transplant Medular din Timisoara intre 2008-2014. Toti pacientii au grefat, prezentand un chimerism cu profil complet de donor precoce post-transplant. In ceea ce priveste complicatiile specifice post-transplant, doi pacienti au prezentat boala grefa contra gazda acuta iar pacientul cu LHH a prezentat boala veno-ocluziva severa. Doi dintre cei trei pacienti cu BGC sunt vindecati, al treilea decedand la 3 luni post-transplant datorita unei pneumonii severe, avand chimerism cu profil 100% de donor si functie granulocitara normala. Unul dintre pacientii cu SCID este bine cu chimerism 100% profil de donor in timp ce fratele geaman a decedat iar pacientul cu LHH este in remisiune. Per global 4 pacienti din 6 sunt in viata, vindecati de imunodeficienta lor severa.

Cuvinte cheie: boala granulomatoasa cronica, limfohistiocitoza hemofagocitica, imunodeficienta severa combinata, transplant de celule stem hematopoietice

HEMATOPOIETIC STEM CELL TRANSPLANTATION FOR PRIMARY IMMUNODEFICIENCIES. THE EXPERIENCE OF THE TRANSPLANT CENTER TIMISOARA

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New achievements could be made for the diagnosis and management of primary immunodeficiencies (PID) and the recent use of exome sequencing technology has led to a more precise genetic characterization of these disorders. Allogeneic hematopoietic stem cell transplantation (HSCT) represents an established indication for several lethal PIDs. Increasing availability of volunteer unrelated donors and more refined HLA typing have led to survival rates reaching 70-80%. We aim to present the experience of the Bone Marrow Transplant Center in Timisoara with HSCT in six pediatric patients with PID. Three children with x-linked chronic granulomatous disease (CGD), one patient with Griscelli syndrome type 2 and hemophagocitic lymphohistiocytosis (HLH) and two other twins with IL-7 receptor deficient T^- B^+ NK^+ severe combined immunodeficiency (SCID) were transplanted from matched related or unrelated donors at our Center between 2008–2014. All of the patients engrafted, presenting complete donor chimerism early after the transplant. Regarding transplant specific complications, two patients presented acute graft versus host disease and the child with HLH suffered from severe veno-occlusive disease early after the transplant. Two out of the three patients with CGD are cured, the third died following a severe pneumonia at 3 months post-transplant with 100% donor chimerism and normal function of granulocytes. One of the twins with SCID is well with 100% donor chimerism and good immune reconstitution, the other one died, whereas the patient with HLH is also in remission. Overall, four of six patients are alive and well with complete recovery of their severe immune deficiency.

Key word: chronic granulomatous disease, hemophagocitic lymphohistiocytosis, severe combined immunodeficiency, hematopoietic stem cell transplantation

BOLI INFECTIOASE / INFECTIOUS DISEASE AND VACCINOLOGY

16. INFECTII PNEUMOCOCICE INVAZIVE LA PACIENTII CU FACTORI DE RISC

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Introducere:

Povara clinica si epidemiologica a bolii pneumococice invazive la copil este inca foarte mare.

In patogenia bolii pneumococice invazive sunt implicate 3 elemente esentiale: colonizarea nazofaringiana , raspunsul inflamator al gazdei , precum si efectul coinfectiilor virale sau bacteriene asupra dezvoltarii pneumococului sau capacitatii de virulenta a acestuia.
Susceptibilitatea cea mai mare o au copiii cu varsta sub 5 ani si in special cei cu varsta sub 2 ani.
In categoria factorilor de risc pentru boala pneumococica invaziva sunt amintite malformatiile congenitale sau traumatismele cu rinolicvoree sau otolicvoree, alaturi de alte afectiuni cronice sau imunodeficiente.
Material si metoda:
Lucrarea actuala pune accent pe malformatii cu fistula LCR ca factor favorizant in determinarea bolii pneumococice invazive de tipul sepsisului sau meningita recurenta. In perioada 2014 -2015 in clinica noastra au fost internate 3 cazuri de sepsis cu meningita cu Streptococcus pneumoniae la copii cu fistula LCR. Prezentam cazul unui pacient in varsta de 4 ani cu sepsis si meningita recurenta cu Streptococcus pneumoniae serotip 6B, rezistent la penicilina,cu interventii repetate pentru fistula nazofrontala cu rinolicvoree.
Concluzii:
Diagnosticul si tratametul acestor afectiuni presupune eforturile considerabile ale unei echipe medicale multidisciplinare avand drept scop recuperarea cat mai rapida a pacientului si evitarea recurentelor.
In perioada 2010-2013 ACIP (Advisory Committee on Immunization Practices) a pubilcat noi recomandari de vaccinare cu PCV 13 si PPSV 23 cu privire in special la pacientii cu factori de risc crescut pentru BPI, subliniind astfel povara clinica inca importanta a acestei afectiuni.

INVASIVE PNEUMOCOCCAL DISEASE IN PATIENTS WITH HIGH RISK FACTORS

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The clinical and epidemiological burden of Invasive Pneumococcal Disease (IPD) is still very increased. In the pathogenesis of Invasive Pneumococcal Disease there are three essential elements involved: nasopharyngeal colonization, the inflammatory response of the host and also the effect of viral and bacterial co-infections on pneumococcus virulence.
The highest susceptibility for IPD is found in children less than 5 years of age, especially in those under 2 years of age. Concerning the risk factors for IPD, the most important are congenital malformations or trauma of the skull with rhinoliquorrhea and otoliquorrhea, along with immunodeficiency and other chronic conditions.
Material and method:
This paper highlights skull malformations with corticospinal fluid (CSF) fistula as a risk factor in developing IPD, like sepsis or recurrent bacterial meningitis. During 2014-2015 in our clinic there have been 3 cases of sepsis with meningitis with Streptococcus pneumoniae in children with CSF fistula. We report the case of a 4-year-old patient with sepsis and recurrent meningitis with Streptococcus pneumoniae serotype 6B, resistant to Penicillin, with repeated surgical interventions for naso-frontal CSF fistula with rhinoliquorrhea.
Conclusions:
The diagnosis and treatment of these affections need considerable effort from a multidisciplinary medical team with the ultimate goal of making a quick recovery of the patient and avoidance of recurrence.
During 2010-2013, ACIP (Advisory Committee on Immunization Practices) published new recommendations for vaccination with PCV13 and PPSV 23 regarding patients with high risk factors for IPD, highlighting the ongoing clinical burden of this affection.

17. CORELATIA DINTRE VIRUSUL HERPES SIMPLEX 1 SI TROMBOZA EXTINSA DE SINUSURI VENOASE CEREBRALE LA UN COPIL CU STATUS PROCOAGULANT

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Introducere
Tromboza de sinusuri venoase cerebrale (TSVC) reprezinta o patologie rara dar deosebit de severa in pediatrie, fiind necesara implicarea unei echipe medicale multidisciplinare in tratarea si dispensarizarea acestor pacienti.
Manifestarile clinice ale TSVC pot fi:cefaleea, varsaturile, fotofobia, tulburari de vedere, convulsii focale sau generalizate, deficite motorii, alterarea statusului mental pana la coma.
Obiectiv: expunerea datelor clinice, paraclinice si de management in tromboza venoasa cerebrala la copil, pe marginea unui caz clinic.
Material şi metoda:

Prezentam cazul unui copil de sex masculin, in varsta de 14 luni, internat in INBI " Matei Bals" pentru febra, varsaturi, convulsii focale si generalizate, sindrom meningean, hemipareza dreapta si coma, simptomatologie neurologica instalata brusc in a 7-a zi de evolutie a unei gingivostomatite herpetice.
RMN cerebral cu angio RMN a evidentiat tromboza extinsa de sinusuri venoase drept, transvers si occipital, cu infarct talamic bilateral, cu transformare hemoragica stanga, hemoragie intraventriculara si hidrocefalie monoventriculara obstructiva.
S-a efectuat profilul protrombotic complet cu depistarea factorilor de risc protrombotic: mutatii de tip heterozigot la nivelul genei MTHFR C677T si PAI -1 4G/5G.
S-a instituit tratament cu Enoxaparina ulterior Acenocumarol, cu evolutie clinica favorabila si recuperare neurologica completa.
Concluzii:
TSVC la copil poate fi complet reversibila cu un diagnostic precoce si cu masuri terapeutice adecvate instituite in timp util.
RMN cerebral cu angio RMN ramane standardul de aur pentru diagnosticarea TSVC.
Profilul protrombotic complet trebuie evectuat la orice copil cu accidente vasculare cerebrale si TSVC.
Virusul herpes simplex poate precipita tromboza la persoanele cu status hipercoagulabil mostenit sau dobandit.
Cuvinte cheie : tromboza de sinusuri venoase cerebrale, angio - RMN, anticoagulant, status hipercoagulabil

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RELATIONSHIP BETWEEN HERPES SIMPLEX VIRUS TYPE 1 AND EXTENSIVE CEREBRAL SINOVENOUS THROMBOSIS IN A CHILD WITH INHERITED HYPERCOAGULABLE STATES

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Background:
Cerebral sinovenous thrombosis (CSVT) is a rare but potentially serious condition in children involving a multidisciplinary team approach .
Clinical manifestations of CSVT include: headache, vomiting, photophobia, blurred vision, focal or generalized seizures, motor deficits, altered mental status and coma.
Objective:
Exposure clinical data, laboratory and management in cerebral venous thrombosis in children- a clinical case report.
Material and method:
We present a case of 14 months - old boy, admitted in ICU National Institute for Infectious diseases "Prof. Matei Bals" for: fever, vomiting, focal and generalized seizures, right hemiparesis and coma. The child was diagnosed with acute herpetic gingivostomatitis one week before onset of neurological symptoms.
MRI brain and angiography revealed extensive thrombosis of the straight, transverse and occipital venous sinuses, bilateral thalamic infarction and left haemorrhagic transformation with intraventricular hemorrhage and univentricular obstructive hydrocephalus.
Thrombophilic screening was performed and a heterozygous mutations genes of C677T MTHFR and 4G/5G PAI was detected.
He was treated with low molecular weight heparin (enoxaparine) followed by oral anticoagulant (acenocumarol) with good clinical outcome and complete neurological recovery.
Conclusions**:**
1. CSVT in children can be fully reversible with early diagnosis and a prompt management.
2. Brain MRI with angio MRI remains the gold standard for diagnosing CSVT.
3. Thrombofilic screening should be considered in any child with strokes and CSVT.
4. Herpes simplex virus may precipitate thrombosis in individuals with inherited or acquired hypercoagulable states.
Key Words
• anticoagulant • cerebral venous sinus thrombosis •magnetic resonance angiography • inherited hypercoagulable states

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18. PARTICULARITATI CLINICE SI BIOLOGICE ALE MENINGITELOR SI ENCEFALITELOR VIRALE

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Infectiile virale ale SNC reprezinta o provocare pentru sectiile de Terapie intensiva ale Clinicilor de Boli infectioase datorita etiologiei variate, ce determina manifestari clinice proteiforme si forme severe de afectare neurologica.
Obiective:

- studiul manifestarilor clinice ale infectiei virale a SNC in functie de virusul implicat.
- identificarea datelor biologice utile in evaluarea gravitatii si a prognosticului in meningo-encefalite virale.

Material si metoda

- Au fost inclusi in studiu pacienti cu varste cuprinse intre 0 – 14 ani, internati in ultimii 3 ani.
- Am urmarit:
  - Variatiile etiologiilor in functie de varsta;
  - Prevalenta semnelor neurologice la debut;
  - Corelatii intre agentul patogen si: modificarile LCR, modificarile imagistice, sechelele neurologice;

Rezultate:

- Au fost cuprinse in studiu 89 de cazuri de infectii virale ale SNC, dintre care 64 au avut etiologie confirmata;
- Prezenta convulsiilor la internare, deficitul motor si sindromul ataxic au o pondere semnificativ statistic crescuta fata de ceilalti factori de risc, in determinarea prezentei sechelelor neurologice.
- Niciunul dintre virusuri nu are un impact semnificativ in determinarea sechelelor neurologice.

Concluzii:

- Aproximativ 27% dintre cazurile de encefalite pot fi prevenite prin vaccinare!
- Chiar atunci cand valorile testelor de rutina ale LCR sunt normale, intensitatea raspunsului inflamator la nivelul SNC este reflectata de „furtuna de citokine” declansata la acest nivel.In toate cazurile prezente in studiul nostru au fost inregistrate valori inalte ale IL-1B, IL-10, TNFα.
- Studiul biomarkerilor din sange si LCR permite: evaluarea severitatii leziunilor cerebrale,monitorizarea progresiei bolii si a eficientei tratamentului.

Cuvinte cheie: meningo-encefalite virale, furtuna de citokine, vaccinare.

CLINICAL MANIFESTATIONS AND BIOLOGICAL PROPERTIES OF VIRAL MENINGITIS AND ENCEPHALITIS

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Viral infections of the CNS is a challenge for intensive therapy of infectious disease clinics due to different etiology, determining various clinical manifestations and severe neurological damage.

Objectives:

- Study of the clinical manifestations of viral infection of the CNS depending on the virus involved.
- Identification of biological data useful in assessing the gravity of viral meningo-encephalitis.

Materials and methods

- Were included in the study patients aged 0-14 years admitted in the last 3 years.
- We look for:
  - Variations by age etiologies;
  - The prevalence of neurological signs at onset;
  - Correlations between pathogen and: CSF changes, changes in imaging, neurological sequelae;

Results:

- Were included in the study 89 cases of CNS viral infections, of which 64 were confirmed etiology;
- Seizure at admission, motor deficit and ataxic syndrome have a significantly increased relative weight of other risk factors in the occurrence of neurological sequelae.
- None of viruses does not have a significant impact in determining neurological sequelae.

Conclusions:

- Approximately 27% of cases of encephalitis can be prevented by vaccination!
- The intensity of the inflammatory response in the CNS is reflected in the "cytokine storm" triggered at this level. In all cases present in our study there were high levels of IL-1B, IL-10, TNF.
- The study of CSF biomarkers in the blood and allows: assessing the severity of brain damage, monitoring disease progression and treatment efficacy.

Key words: viral meningo-encephalitis, cytokine storm, vaccination

19. COLITA CU CLOSTRIDIUM DIFFICILE LA COPIL – FORME CLINICO-EVOLUTIVE SI TRATAMENT

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Introducere
Infectia cu *Clostridium difficile* la copil, in functie de varsta si de statusul imun, imbraca diverse forme clinice de boala, de la asimptomatic la colita pseudomembranoasa, severa. Frecvent colita cu *Clostridium difficile* este o infectie nosocomiala.
Obiective
In aceaste lucrare ne-am propus sa analizam formele clinico-evolutive ale colitei cu *Clostridium difficile* la copil.
Material si metoda
Am efectuat un studiu retrospectiv, in perioada 2010 – 2014, asupra cazurilor de colita cu Clostridium difficile la copil, internate in Institutul National de Boli Infectioase ”Prof. dr. Matei Bals” Bucuresti. La aceste cazuri am urmarit o serie de parametrii (varsta, sex, statusul imun), forma clinica de boala precum si tratamentul administrat. Diagnosticul etiologic a fost stabilit prin coprocultura, PCR pentru clostridium toxigen din materiile fecale.
Rezultate
In perioada analizata am inregistrat 20 cazuri de colita cu *Clostridium difficile* la copil. 25 % erau pacienti cu imunodepresie (3 copii cu afectiuni maligne pentru care au primit chimioterapie si 2 cazuri de infectie HIV). Cazurile predomina la grupa de varsta 4 – 8 ani, la sexul feminin. Toti pacientii au primit tratament conform protocolului international de tratament. 15 % dintre cazuri au prezentat o recidiva sau doua, iar 10 % mai mult de 3 recaderi motiv pentru care s-a efectuat recolonizare bacteriana cu microbionta de la donator sanatos. Toate cazurile au evoluat favorabil. Nu am inregistrat decese. Cele doua cazuri la care s-a efectuat recolonizare bacteriana nu au mai prezentat recidiva.
Concluzii
Infectia acuta cu *Clostridium difficile* la copil, mai ales la imunodeprimati poate imbraca forme clinico-evolutive severe. In cazul recidivelor multiple recolonizarea intestinala bacteriana poate fi o solutie de succes.

CLOSTRIDIUM DIFFICILE COLITIS IN CHILDREN – CLINICAL FORMS AND TREATMENT

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Introduction
The pediatric cases of Clostridium difficile infection (CDI), based on the host immune status, can present under a broad spectrum of clinical forms, from asymptomatic portage of the germ up to severe, life-threatening cases of pseudo-membranousous colitis.
Objective
In this work we aimed to analyze the clinical forms and treatment of Clostridium difficile colitis in children.
Material and method
We conducted a clinic-based retrospective surveillance witch analyzed the cases of confirmed CDI in infants and children that were treated and monitored at The National Institute for Infectious Diseases "Prof. Dr. Matei Bals", Bucharest during the period 2010 – 2014. For the selected cases the clinical and demographic patient features followed were: age, sex, immune status, severity of the disease and the treatment regimen. The etiological diagnosis was made by culturing the stool samples and by PCR testing for Toxigenic *Clostridium difficile strains.*
Results:
During the survey period we registered 20 cases of CDI in infants and children. On subsets, 25 % of the cases were pediatric immunodepresed. Based on the demographic features we observed a female predominance and a pick of incidence in the 4-8 years subgroup. All the diagnosed patients were treated according to the international CDI treatment protocol. We registered a 15 % recurences rate, with one or two relapsing episodes. One tenth of our cases presended more than 3 CDI relapsing episodes and consequently received a fecal microbiota transplantation with the transplanted material being donated by a healthy family member, with subsequent full recovery. The cases that underwent the stool transplant protocole did not registerd relapsing episodes. No fatal cases were registered.
Conclusions:
The CID in infants and children, especially in imuno-compromised hosts, can present under sever clinical forms. In the case of multiple relapsing episodes the fecal microbiota transplantation procedure can be a successful treatment option.

20. LOCUL INFECTIEI CU BORDETELLA SPP. IN RANDUL PATOLOGIEI RESPIRATORII DOMINATE DE TUSE, ANALIZA RETROSPECTIVA A CAZURILOR PEDIATRICE DIN PERIOADA 2010-2014.

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Introducere:
Tusea reprezinta simptomul caracteristic al patologiei respiratorii in randul copiilor si este unul din principalele motive pentru prezentarea la medic.
Material si Metoda:
Am analizat, intr-o maniera retrospectiva, evolutia pacientilor internati in departamentul pediatric al INBI "Matei Bals" – Bucuresti, in perioada 2010 – 2014 care au prezentat tuse cu o durata de cel putin o saptamana la care au asociat unul dintre urmatoarele 3 simptome: accese paroxistice de tuse; inspir zgomotos ("whooping"); voma post-tuse. Pentru cele 790 de cazuri selectionate am urmarit caracteristicile epidemiologice si demografice ale grupului, forma de boala si evolutia sub tratament. Incadrarea etiologica a fost stabilita pe baza testelor serologice
Rezultate :
Din cele 790 cazuri selectionale 13,8 %, dintre care mai mult de jumatate nevaccinati DTP, au avut ca agent etiologic Bordetella Spp, reprezentand 11% din cazurile de Tuse convulsive raportate national pe perioada celor 5 ani de supraveghere. Restul de 682 de cazuri au avut ca agent etiologic Virusul Sincitial Respirator (39,7%), Adenovirus (21,5%), Mycoplasma Pneumoniae (18,3%), Chlamydia Pneumoniae (6,7% din cazuri).
Forme severe de boala au imbracat 279 dintre cazuri si au necesitat in medie 3 zile de monitarizare in serviciul de terapie intensiva. Perioda medie de spitalizare inregistrata a fost de 6,9 zile. Toate complicatiile severe au fost inregistrate la copii cu varsta <6 luni. Nu s-au inregistrat decese la nivelul cohortei studiate pe perioada de monitorizare.
Concluzii:
Tusea convulsiva ramane endemic in Romania.
Cuvinte cheie: tuse convulsiva; Bordetella Spp;

THE PLACE OF THE BORDETELLA SPP. INFECTION IN THE RESPIRATORY SINDROMS DOMINATED BY COUGH IN INFANTS AND CHILDREN, A 5 YEARS SURVEY

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Introduction
Cough is a common indication of respiratory illness and is one of the more common symptoms of children seeking medical attention.
Material and Method.
We conducted a clinic-based retrospective surveillance witch analyzed the cases managed at "The National Institute for Infectious Diseases "Prof. Dr Matei Bals" – Pediatric Department, Bucharest during the period of January 2010 up to December 2014 who presented accusing couch episodes from over a week and who associated one of the following symptoms: paroxysms of coughing, inspiratory "whoop," posttussive vomiting or apnea. We selected 790 suitable cases for whom we analyzed: age, sex, vaccinal status, severity of the disease and the complications. The etiological diagnosis was made by serologic testing for Bordetella, Mycoplasma, Chlamydia , Adenovirus and by rapid testing for Sincitial Respiratory Virus (RSV).
Results and findings.
Based on the etiological stratification 108 patients (13,8 %) were diagnosed with Bordetella Spp infections, 62,4% of them being completely unvaccinated against Pertusis, representing 11% of the national reported cases of Whooping Cough during the 5 years of survey. With decresing frequencies the rest of the cases (682) were caused by: RSV (39,7%), Adenovirus (21,5%), Mycoplasma (18,3%), Chlamydia (6,7%).
The majority of the cases evolved favorable, no fatal cases were registered but 279 presented with initial altered status and required, on average, 3 days of Intensive Care Unit management. The mean hospitalization period registered is 6,9 days. All the severe complications were registered in the < 6 months age group.
Conclusion.
Whooping cough remains endemic in Romania.
Key words: whooping cough; Bordetella Spp.

NEFROLOGIE/ NEPHROLOGY

21. SINDROMUL NEFROTIC - UTILITATEA TESTARII GENETICE LA COPII

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Introducere: Sindromul nefrotic (SN) este cauzat de diferite afectiuni care se soldeaza cu distructia rinichilor. Studii recente au descris deja mutatii implicate in etiologia, prognosticul si raspunsul la terapie.
Scop: S-a cercetat existenta unei asocieri a unor polimorfisme genice (ACE I/D; VEGF, NPHS2 r229Q; eNOS 4a/bVNTR;TNF alfa G308A, IL174G>C) si SN.
Material si metoda: Studiul a cuprins 57 pacienti cu SN (congenital, idiopatic) si un grup de 123 copii sanatosi. S-a utilizat metoda PCR sau ARMS-PCR folosind primeri adecvati.
Rezultate: Mutatii in gena NPHS2 r229Q si TNF alfa s-au observat doar la cazurile de SN congenital. S-a observat o incidenta mai mare, a genotipului II si ID la pacientii cu SN versus martor (p>0.05) mai ales la sexul feminin. Nu s-a observat diferente in ceea ce priveste mutatia genei eNOS 4a/b VNTR intre cele 2 loturi
Concluzii: In studiul de fata nu am putut demonstra existenta unei asocieri intre polimorfismul genei eNOS4a/b VNTR; IL-6 174G>C gsi sindromul nefrotic. S-a observat o asociere intre SN si polimorfismul ACE I/D, I reprezentand un factor de risc in aparitia SN. Mutatile genei VEGF au rol protector in dezvoltarea SN.Mutatia in gena NPHS2 r229Q si TNF alfa reprezinta un factor de risc in aparitia SN congenital. Sunt necesare studii suplimentare cu un numar mai mare de pacienti pentru a clarifica asocierea genelor studiate si SN.
Acknowledgments: Aceasta lucrare a fost suportata de Grantul Intern de Cercetare a Universitatii de Medicina si Farmacie Tîrgu Mureş, Romania, contract nr 29/11.12.2013

NEPHROTIC SYNDROME - UTILITY OF GENETIC TESTING IN CHILDREN

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Background: Nephrotic syndrome (NS) is caused by different disorders that damage the kidneys. Recent studies have described that certain genetic mutations are involved in the NS pathway, prognostic and response to therapy.
Aim: This study was performed to investigate if there is any association of some polymorphisms (ACE angiotensin converting enzyme I/D gene polymorphism,VEGF (vascular endotelial growth factor), NPHS2, eNOS4a/b VNTR (nitric oxide sintetase) , Tumor Necrosis Factor α (TNF α) G308A and IL-6 174G>C gene polymorphism and nephrotic syndrome.
Materials and methods: Our study included a number of 57 patients with NS and a control group which included 123 healthy persons. The gene polymorphisms were determined by the PCR and PCR-ARMS techniques using specific primers.
Results: Mutations (NPHS2 and TNF alfa) were detected in 100% of congenital-onset NS. Patients with NS had a higher percentage of II and ID genotype than the control group (especially in girls).
Conclusions: We were unable to show a relationship between eNOS4a/b VNTR; IL-6 174G>C gene polymorphism and NS. There is an association beetwen NS and ACE I/D polymorphism, the deletion might be a protective factor and insertion a risk factor. Mutation in NPHS2 and TNF alfa gene represent a risk factor for congenital NS.
Further studies with larger samples are needed to be done to clarify any association between these polymorphisms and NS.
Acknowledgments: This paper work was supported by Internal Research Grants of the University of Medicine and Pharmacy Tîrgu Mureş, Romania, contract no 29/11.12.2013
Key words: children, nephrotic syndrome, gene polymorphisms

22. PARTICULARITATI DE EVOLUTIE IN SINDROMUL HEMOLITIC SI UREMIC - STUDIU RETROSPECTIV PE 5 ANI

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Post-*diarrheal* Haemolytic Uraemic Syndrome (HUS D+) is common cause of acute renal injury in childhood. The aim is to find predictors of severe evolution of HUS. Descriptive, retrospective study, carried out in our clinic, from Jan 2010–Dec 20114. We defined HUS - hemolytic anemia, thrombocytopenia and acute kidney injury (AKI). Monitored demographics dates, clinical, laboratory parameters, evolution, treatment. RESULTS: 62 children 4-192 monts (mo), diagnosed with HUS D +, between 2010-2014. 71% - age <18 mo, 75% boys. Mean follow-up 24 mo. Anuria was present in 75% cases. Faster recovery of renal function (17d) was in patients in whom dialysis was initiated in the first 24h. 55% patients needed dialysis. Neurological phenomena was in 20%cases. 66% had hypertension (HTA), in 13 patients complicated with lung edema and strokes. 35%patients had leukocytes >20.000, 27% severe thrombocytopenia<30.000. The

combination of the two established a strong correlation (p <0.01) with renal during recovery. All patients associated anemia, who correlates directly, poorly with renal function recovery. Persistent proteinuria after one year was identified in 31%cases. Unfavorable evolutions to death occurred in 7% cases. All patients who died were aged<18 mo, associated late presentation, long anuria, neurological phenomena, HTA, leukocytosis, severe thrombocytopenia, infectious complications. CONCLUSION: HUS is important AKI cause in children. Over 50% patients requiring dialysis. Bad prognostic factors are leukocytosis, thrombocytopenia, duration of anuria, neurological phenomena, severe hypertension. Persistent proteinuria at 1 year is a predictor of unfavorable evolution. Rapid recognition increase the chance of complete recovery of renal function.

23. MANAGEMENTUL HIDRONEFROZEI DEPISTATE ANTENATAL – EXCES DE INVESTIGATIE VS. INTARZIERE IN DIAGNOSTIC

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Posibilitatea remisiunii spontane a hidronefrozei antenatale (HAN) este o realitate dovedita. Scopul studiului consta in identificarea incidentei uropatiilor semnificative clinic la pacientii cu HAN pentru realizarea unui protocol postnatal adaptat care să permită un diagnostic precoce şi să evite investigaţii şi tratamente inutile. Material, metoda: analiza retrospectiva a 106 copii cu HAN evaluati postnatal in Clinica IV Nefrologie Pediatrica Iasi in perioada 2010 - 2014. In functie de gradul de hidronefroza la ecografia postnatala (clasificarea Societatii de Urologie Fetala) am format 2 loturi : A - hidronefroza usoara (gradele1,2), B - hidronefroza medie si severa (3,4) ureterohidronefroza, sau malformatie complexa. Am urmarit comparativ: tipurile de malformatii, investigatiile radiologice, profilaxia antibiotica, corelate cu aparitia infectiei urinare (ITU) si necesitatea chirurgiei. Rezultate : 42,5% din cazuri au fost incadrate ca hidronefroza izolata de grad mic, urmate de viciul de jonctiune pieloureterala (23,5%) si refluxul vezicoureteral (RVU) 15 %. La lotul A (48 cazuri) s-au efectuat examene radiologice la 23% cazuri si 2% au avut ITU, comparativ cu lotul B (54 cazuri) cu 89% cazuri investigati radiologic, 15% cu ITU si 61% operati. Concluzii. 31% din pacientii cu HAN au prezentat uropatii semnificative clinic, 96 % apartinand lotului B. In protocolul de urmarire postnatala este suficienta scanarea ecografica seriata a hidronefrozei usoare. Profilaxia antibiotica trebuie utilizata la pacientii cu hidronefroza moderata si severa, cei cu ureterohidronefroza, la pacientii simptomatici (ITU) si in RVU. Chirurgia trebuie rezervata malformatiilor obstructive si in RVU de grad mare persistent dupa varsta de 1 an.

ANTENATAL HYDRONEPHROSIS MANAGEMENT – OVERINVESTIGATIONS VS. DIAGNOSTIC DELAY

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The spontaneous remision of antenatal hydronephrosis (AHN) is a documented posibility. The aim of the study is identification of clinically significant uropathy incidence and establish a postnatal follow-up and management protocole for early diagnosis and avoid usefulness investigations and treatments. Method: retrospective study of 106 children with AHN followed postnatally in the Pediatric Nephrology Department of our hospital between 2010-2014. The children were divided in 2 groups based on the postnatally ultrasound hydronephrosis degree (Fetal Urology Society classification): A – mild hydronephrosis (grade 1,2), B – medium and severe hydronephrosis (grade 3,4) or ureterohydronephrosis, or complex malformation. We analysed: malformation type, radiological exams, antibiotic prophylaxis, correlated with urinary tract infection (UTI) and the need of surgery. Results. 42.5% cases were classified as isolated hydronephrosis of mild degree, followed by pieloureteral obstruction (23.5%) and vesicoureteral reflux (VUR) 15%. In group A (48 cases) 23% had radiological exam, 2% had UTI, comparing with group B (54 cases) with 89% radiological exam, 15% UTI and 61% surgery. Conclusions. 31% of the patients with AHN had clinicaly significant uropathy, 96% from group B. In postnatal follow up protocol mild hydronephrosis can be followed through seriated ultrasound studies. Antibiotic prophylaxis must be used in moderate and severe hydronephrosis, ureterohydronephrosis, UTI and VUR. Surgery is reserved for obstructive malformation, persistent severe VUR after 1 year.

24. NEFROPATII ASSOCIATE INFECTIEI HIV

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Scop-evidenţierea tipurilor de afectare renală apărute în cadrul infecţiei cu HIV/SIDA.

Introducere Pe măsură ce speranţa de viaţă a pacienţilor a crescut, datorită eficienţei HAART, afectarea renală asociată acestei infecţii a devenit o problemă importantă.

Material/metoda Este un studiu retrospectiv pe 202 pacienţi cu afectare renală, dintr-un total de 640 de pacienţi cu infecţie HIV/SIDA monitorizaţi în Clinica III Pediatrie Timişoara în perioada 1990-2013.

Rezultate şi discuţii Majoritatea sunt născuţi în perioada 1988-1990 (88,61%). Cale de transmitere -necunoscută (70,9%), sânge (8,91%), gamaglobulina administrată im (7,92%), nosocomială (7,92%), verticală (2,47%), prin intermediul altor

produse de sânge (1,98%). O treime dintre pacienţii cu infecţie HIV/SIDA au avut manifestări renale. 152 cazuri au fost cu infecţii urinare joase (74,24%) şi 17 au avut infecţii urinare înalte (8,41%). 17 pacienţi au avut litiază renală sau episoade de colică, 6 au fost cu enurezis, 3 cu insuficienţă renală cronică,3 cu insuficienţă acută, 3 au avut hidronefroză. Nivelul CD4+ în momentul diagnosticării infecţiei HIV/SIDA a arătat faptul că aproape jumătate dintre pacienţi (47,52%) pot fi consideraţi „*late presenter"* fiind cu o valoare a limfocitelor T CD4 sub 200. Nefropatia asociata infectie HIV (HIVAN) are mecanisme fiziopatologice variate: imunodeficienţa asociată, medicaţia administrate si comorbidităţile (hipertensiunea arterială, diabetul zaharat, hepatita).
Concluzii Distribuţia în funcţie de vârsta pacienţilor în momentul diagnosticării infecţiei a fost în concordanţă cu datele din literatură, dar afectarea renala pare sa fie mai redusa (mai ales fata de populatia afro-americana). Am descoperit efectuand punctie biopsie renala 2 cazuri ce pot fi incadrate ca HIVAN.

HIV-ASSOCIATED NEPHROPATHY

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Aim – to highlighte the types of renal dysfunction occurred in HIV / AIDS.
Introduction
As the life expectancy of patients has increased due to HAART, the kidney damage associated with this infection has become an important issue.
Material / method
This is a retrospective study of 202 patients with renal impairment, out of a total of 640 patients with HIV / AIDS monitored Timisoara (3rd pediatric clinic) during 1990-2013.
Results and discussion
Most are born between 1988-1990 (88.61%). Transmission path - unknown (70.9%), blood (8.91%), gamma globulin administered im (7.92%), nosocomial (7.92%), vertically (2.47%) through other blood products (1.98%). A third of patients with HIV / AIDS had renal manifestations. 152 cases were lower urinary tract infections (74.24%) and 17 had high urinary tract infections (8.41%). 17 patients had kidney stones or colic episodes, 6 were with enuresis, three with chronic renal failure, three with acute renal injury, 3 had hydronephrosis. CD4 + levels at diagnosis moment of HIV / AIDS has shown that nearly half of patients (47.52%) can be considered "late presenter '- a value below 200 CD4 T lymphocytes. HIV-associated nephropathy (HIVAN) has variated pathophysiological mechanisms: immunodeficiency, associated comorbidities and medication administered.
Conclusion
Distribution by patient age at diagnosis of infection was consistent comparative with the literature, but kidney damage appears to be lower (especially to the African-American). We found, performing renal biopsy, 2 cases that can be classified as HIVAN.

NEONATOLOGIE/ NEONATOLOGY

25. CHIST BRONHOGEN CONGENITAL – CAUZĂ DE DETRESĂ RESPIRATORIE LA NOU-NĂSCUT

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Introducere: Detresa respiratorie neonatală reprezintă orice dificultate respiratorie în primele 28 zile de viaţă. Chistul bronhogen congenital este o malformaţie pulmonară rară care se poate manifesta în perioada neonatală cu detresă respiratorie de grade diferite şi care necesită o abordare diagnostică şi terapeutică promptă. Material şi metodă: autorii prezintă un caz de chist bronhogen diagnosticat în Centrul Regional de Terapie Intensivă Neonatală Tg. Mureş. Rezultate: Un nou-născut prematur, cu vârsta de gestaţie de 36 săptămâni, sex masculin, cu greutatea la naştere 2600g, prezintă la 24 ore postpartum detresă respiratorie manifestată prin geamat expirator, polipnee, tiraj subcostal şi intercostal moderat, cianoză responsivă la oxigenoterapie, torace bombat. La VG de 20 săptămâni se decelează ecografic o formaţiune chistică în lobul mijlociu al plămânului drept fetal. Radiografia toracică şi computer tomografia evidenţiază chist aeric bazal drept anterior cu deplasarea mediastinului spre stânga. Analiza gazelor arteriale evidenţiază hipoxemie, hipercarbie şi acidoză respiratorie. Se intervine

chirurgical efectuâdu-se chistectomie, cu evoluţie favorabilă. Concluzie: malformaţiile congenitale pulmonare, ridică probleme de diagnostic diferenţial care trebuie rezolvate în vederea stabilirii unei atitudini terapeutice corecte.
Cuvinte cheie: chist bronhogen, detresă respiratorie neonatală

CONGENITAL BRONCHOGENIC CYST – CAUSE OF RESPIRATORY DISTRESS IN NEONATES

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Introduction: the neonatal respiratory distress means any breathing difficulty in the first 28 days of life. Congenital bronchogenic cyst malformation is a rare lung malformation that can occur in the neonatal period with varying degrees of respiratory distress and require prompt diagnostic and therapeutic approach.
Material and method: The authors present a case of bronchogenic cyst diagnosed in Regional Center NICU Tg. Mures.
Case presentation: A premature male newborn at 36 weeks gestational age, birth weight 2600g, developed at 24 hours of age respiratory distress manifested by grunting, tachypnea, moderate subcostal and intercostal retractions, cyanosis responsive to oxygen therapy, bulging chest. At 20 weeks of gestational age, ultrasound examination revealed a cystic formation in the middle lobe of the fetal right lung. Chest X-ray and computer tomography examinations reveal right anterior basal cyst, air leakage with mediastinal shift to the left. Blood gas analysis showed hypoxemia, hypercarbia and respiratory acidosis. Cystectomy was performed by thoracotomy, with favorable outcome.
Conclusion: Congenital lung malformations require differential diagnosis in order to establish the proper terapeutic options.
Keywords: bronchogenic cyst, neonatal respiratory distress

26. AFECŢIUNI NEUROLOGICE PERINATALE CU RISC MAJOR PENTRU PROGNOSTIC NEURODEVELOPMENTAL NEFAVORABIL – INCIDENŢĂ ŞI CORELAŢII CLINICO-EPIDEMIOLOGICE LA PREMATURUL CU VÂRSTĂ DE GESTAŢIE SUB 32 DE SĂPTĂMÂNI

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Complicaţii ale prematurităţii precum hemoragie intraventriculară severă (HIV)(grad 3-4) şi leucomalacia periventriculară (LPV) reprezintă factori de risc major pentru un prognostic neurodevelopmental nefavorabil pe termen îndelungat.
Scop: Autorii şi-au propus să evalueze incidenţa şi corelaţiile clinică-epidemiologice ale HIV severe şi LPV la prematurii cu vârstă de gestaţie (VG) sub 32 de săptămâni.
Material şi metodă: Studiul prospectiv s-a desfăşurat pe o perioadă de 2 ani (01.01.2010-21.12.2011) şi se bazează pe datele colectate în Registrul Naţional de Detresă Respiratorie (SDR). Au fost incluşi în studiu toţi prematurii cu VG≤32 săptămâni iar analiza datelor a fost efectuată cu IBM SPSS Statistics 19 (p<0,05, CI 95%)
Rezultate: Lotul de studiu a cuprins 139 prematuri cu VG medie de 30,3±1,9 săptămâni şi greutate medie la naştere (GN) de 1412,9±367,9g. HIV severă a fost identificată în 7 cazuri (5,3%) iar LPV în 5 cazuri (3,5%). În timp ce prezenţa HIV severă s-a asociat statistic semnificativ cu GN şi VG mici (p=0,000), scor Apgar mic la 1 minut (p=0,023), naşterea în maternitate de nivel inferior (p=0,020) şi SDR sever (relevat de nevoia administrării de surfactant, de ventilaţie asistată şi persistenţa canalului arterial – p 0,001-0,003; OR 8,9-11,9) prezenţa LPV s-a corelat semnificativ cu administrarea antenatală de cortizon (p=0,026), durata prelungită a suportului respirator de tip CPAP (p=0,037) şi apneea prematurului (p=0,007; OR 9,3).
Concluzie: Deşi în mare măsură rezultatele analizei informaţiilor colectate sunt asemănătoare datelor raportate în literatura de specialitate numărul mic de cazuri de HIV şi LPV (ceea ce este îmbucurător) limitează valoarea semnificaţiei acestora.
Cuvinte cheie: prematuritate, hemoragie intraventriculară, leucomalacie periventriculară, factori de risc, epidemiologie

PERNATAL NEUROLOGICAL CONDITIONS WITH MAJOR RISK FOR UNFAVORABLE NEURODEVELOPMENTAL OUTCOME – INCIDENCE, CLINICAL AND EPIDEMIOLOGICAL CORRELATIONS IN PRETERM INFANTS LESS THAN 32 WEEKS GESTATION

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Complications of prematurity as severe intraventricular hemorrhage (IVH) (grade 3-4) and periventricular leukomalacia (PVL) are major risk factors for an unfavorable neurodevelopmental long term outcome.
Aim: The authors proposed theselves to evaluate the incidence, clinical and epidemiological correlations of severe IVH and PVL in preterm infants less than 32 weeks gestation.

Material and methods: The prospective study was developed over a 2 years period (01.01.2010 - 31.12.2011) and is based on the information collected in the National Registry for Respiratory Distress Syndrome (RDS). All preterm infants ≤32 weeks gestational age were included and data analysis was performed using IBM SPSS Statistics 19 (p<0.05, 95% CI).
Results: The study group comprised 139 preterm infants with gestational age (GA) of 30.3±1.9 weeks, and mean birth weight (BW) of 1412.9±367.9g. Severe IVH occurred in in 7 cases (5.3%) and PVL in 5 cases (3.5%). While severe IVH was significantly associated with lower GA and BW (p=0.000), low Apgar score at 1 minute (p=0.023), birth in a lower level unit (p=0.020), and severe RDS (revealed by need for surfactant administration, mechanical ventilation, and persistent ductus arteriosus – p 0.001-0,003; OR 8.9-11.9) PVL occurrence was significantly correlated with antenatal corticosteroid administration (p=0.026), prolonged CPAP respiratory support (p=0.037), and apnea of prematurity (p=0.007; OR 9.3).
Conclusion: Event though in a great measure the results from the collected data analysis are resembling the data reported in the literature, the low number of IVH and PVL cases (which is a good news) is limiting the value of their significance.
Key words: prematurity, intraventricular hemorrhage, periventricular leukomalacia, risk factors, epidemiology

27. INFECȚIA PERINATALĂ CU STREPTOCOC GRUP B – FACTORI DE RISC SI ASPECTE CLINICE şI PARACLINICE

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Streptococul degrup B este factor de risc pentru infectia perinatala.
Lipsa specificitatii semnelor clinice, numărul mare de cazuri infraclinice (nou-nascuti aparent sanatosi) pot intarzia diagnosticul si initierea tratamentului infectiei perinatale.
Obiective, scop.
Stabilirea incidentei colonizării materne si neonatale cu *Streptococ de grup B,* identificarea factorilor de risc pentru infectie perinatala cu *Streptococ de grup* B, depistarea precoce a acesteia cu scopul diminuarii morbiditatii si mortalitatii neonatale.
Material si metoda.
Studiu efectuat in Polizu, pe o perioadă de 7 ani (2006-2012) cuprinzand 24521 cupluri mama-nou-nascut. Corelatiile referitoare la factori de risc, date clinice si paraclinice s-au efectuat analizand 125 nou-nascuti colonizati cu *Streptococ grup B* care au evoluat cu si fara infectie precoce. Datele au fost prelucrate statistic.
Rezultate.
Streptococ grup B a fost depistat la 5,79% mame. 12,09% din nou-nascutii acestora au fost si ei colonizati. 4,21% nou-nascuti colonizati au provenit din mame necolonizate.
Din 125 nou-nascuti colonizati cu *Streptococ grup B,* doar 1,6% au evoluat cu infectie severa (septicemie, meningita). Semne clinice initiale, evocatoare de infectie (debut in 60% cazuri in primele 12 ore de viata) frecvente au fost meteorism abdominal (39,13%) geamat expirator (34,78%) tiraj intercostal (30,43%) manifestari cutanate (marmorare, cianoza), edeme (21,73%). Exista diferente semnificative statistic intre nou nascuti cu si fara infectie, privind factorii de risc asociati (asfixie perinatala), semnele clinice si de laborator.
Concluzii. Cunoasterea factorilor de risc (mai mare in asocierea mai multor factori pentru acelaşi caz), a semnelor clinice de debut, pentru depistarea precoce a infectiei perinatale, pot diminua morbiditatea neonatala.

GROUP B STREPTOCOCCUS PERINATAL INFECTION – RISK FACTORS, CLINICAL AND PARACLINICAL FACTS

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Introduction (Background) *Group B Streptococcus* represents a frequent risk factor for perinatal infections. The lack of specific clinical findings, the numerous sub-clinical infections (apparently healthy newborns) may delay the moment the of diagnosis and the beginning of the treatment.
Objective To assess the incidence of both maternal and newborn colonisation by *Group B Streptococcus*, to identify risk factors for peri-natal *Group B Streptococcus* infection and early diagnosis of the infection in order to lower the neonatal morbidity and mortality rates.
Material and methods Retrospective study on 24521 cases (mother / child pairs) from „Polizu Hospital", during 2006 and 2012 (7 years). Correlation between risk factors, clinical and para-clinical data were targeting 125 *Group B Streptococcus* colonised newborns, with and without early onset sepsis.
Results *Group B Streptococcus* was detected in 5,39% of the mothers and in12,09% of their newborns. 4,21% of the colonised newborns had *Group B Streptococcus* negative mothers. From the 125 colonised babies only 1,6% developed a severe infection (sepsis, meningitis). Frequent initial clinical signs pointing an infection (onset during first 12 hour of life in

60% of cases) were abdominal distension (39,13%), grunting (34,78%), chest retraction (30,43%), discoloration (mottling, cyanosis) or oedema (21,73%). Between the healthy and the infected newborn groups statistically significant differences were found, regarding the coexisting risk factors (perinatal asphyxia), clinical signs and paraclinical data.
Conclusions. Identifying risk factors and pinpointing the augmentation of risk with their cumulation, determining clinical signs at onset and criteria for precocious diagnosis may lower neonatal morbidity.

28. ANTIBIOREZISTENŢA GERMENILOR GRAM NEGATIVI – SEMNAL DE ALARMĂ PENTRU SECŢIILE DE TERAPIE INTENSIVĂ NEONATALĂ

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Introducere: În secţiile de terapie intensivă neonatală antibioticele, cele mai eficiente instrumente de luptă împotriva patogenilor, sunt adesea utilizate excesiv, consecinţa fiind selectarea de tulpini multidrogrezistente.
Obiectiv: analiza rezistentei la antibiotice a bacililor gram-negativi intr-o sectie de neonatologie de nivel III din Romania, în perioada 2009-2013.
Material şi metodă: am realizat un studiu retrospectiv bazat pe analiza culturilor periferice, de pe cateter venos central şi a hemoculturilor pe o perioada de 5 ani (2009-2013) în vederea evaluării sensibilităţii/rezistenţei la antibiotice.
Rezultate: Germenii gram-negativi izolaţi cel mai frecvent la nou-născuţii din unitatea noastră au fost specii de *E. coli*, *Klebsiella*, *Entetobacter*, *Serratia* şi *Acinetobacter*. Analiza statistică demonstrează o scădere semnificativă ($\chi 2=79.2$) a sensibilităţii la antibiotice a germenilor gram-negativi în anul 2013 comparativ cu anul 2009, aspect demonstrat de corelaţia ($r=-0.72$, $p<<0.01$, CI de 95%) între sensibilitate şi perioada studiată. Aprecierile au fost făcute pentru un interval de confidenţă de 95%.
Concluzii: Practicile medicale curente şi utilizarea extensivă a antibioticelor cu spectru larg au contribuit la selectarea de tulpini multirezistente. Utilizarea restrânsă a antibioticelor este tot mai mult promovată ca o metodă de limitare a antibiorezistenţei multiple, de îmbunătăţire a calităţii îngrijirilor şi de evitare a complicaţiilor infecţioase.

ANTIMICROBIAL RESISTANCE OF GRAM NEGATIVE MICROORGANISMS – A DISTRESS SIGNAL FOR NEONATAL INTENSIVE CARE UNITS

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Introduction: In neonatal intensive care units all over the world antibiotics, the most efficient tools against pathogenic microorganisms are often used excessively, resulting in selection of multidrug resistant strains.
Aim of study: Analysis of resistance to antibiotics of gram negative bacteria isolated over a period of five years (2009-2013), in a level III neonatal unit in Romania.
Material and method: We performed a retrospective study based on the analysis of peripheral, central vein catheters and blood cultures sampled from newborns admitted in the Neonatology Department of ”Cuza-Voda” Clinical Hospital of Obstetrics and Gynecology of Iasi between 2009 and 2013, in order to evaluate the sensitivity/resistance to antibiotics of the gram negative microorganisms.
Results: The gram negative pathogens most commonly isolated from peripheral and central cultures were *E. coli*, *Klebsiella*, *Enterobacter*, *Serratia* and *Acinetobacter* species. The statistical analysis showed a significant reduction ($\chi 2=79.2$) of the sensitivity to antibiotics of gram negative microorganisms in 2013 in comparison with the year 2009, as proved by the correlation ($r=-0.72$, $p<<0.01$, 95% CI) between the sensitivity and the period studied. The assessment was performed for a confidence interval of 95%.
Conclusions: Current medical practices and excessive empirical use of broad spectrum antibiotics play a massive role in the selection of multidrug resistant pathogens. Restrictive policies for the empirical use of antimicrobial agents should be promoted as a means to prevent the appearance of multidrug resistance and infectious complications, and also to improve the quality of care.

NEUROLOGIE SI PSIHIATRIE INFANTILA / CHILD NEUROLOGY AND PSYCHIATRY

29. EVALUAREA SI INTERVENTIA ONLINE IN TULBURARILE DE ANXIETATE LA ADOLESCENTI

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In ultimii ani, datorita accesului relativ facil la resurse informationale adecvate sau campaniilor de destigmatizare a tulburarilor psihice, solicitarea serviciilor specializate de psihiatrie pediatrica a crescut considerabil. In acelasi timp, numarul de instrumente disponibile de screening sau evaluare, precum si posibilitatile de interventie, au devenit din ce in ce mai variate. Cu toate acestea, utilizarea algoritmilor de diagnostic recomandati de experti sau a terapiilor evidence based este de cele mai multe ori restrictionata. Acest fapt poate fi explicat, in contextul cresterii numarului pacienti, de resursele umane sau financiare insuficiente pentru a oferi servicii de calitate optima intr-un timp scazut. In vederea implementarii si utilizarii eficiente a unei platformei web destinate screeningului, evaluarii si terapiei tulburarilor anxioase ale copiilor si adolescentilor am aplicat un chestionar adresat medicilor psihiatri. S-au urmarit in acest fel realizarea unei imagini de ansamblu asupra practicilor actuale in tulburarile anxioase (diagnostic, terapie, management al cazului) din serviciile specializate de psihiatrie pediatrica din tara. Astfel, prin aplicarea chestionarului, am incercat pe de o parte evaluarea metodelor actuale de interventie utilizate in practica curenta de catre pedopsihiatri, si pe alta parte, evaluarea perceptiei si disponibilitatii acestora de a folosi evaluarea si interventia online.

platforma online, evaluare, interventie, psihiatrie pediatrica

ONLINE ASSESSMENT AND INTERVENTION IN ADOLESCENT ANXIETY DISORDERS

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In recent years the request for specialized pediatric psychiatry services has increased considerably due to relatively easy access to appropriate information and the de-stigmatization campaigns around mental disorders.At the same time, the available tools for screening or evaluation, and intervention possibilities have become increasingly diverse.However, the use of diagnostic algorithms recommended by experts and evidence based therapies is often restricted.This can be explained, in view of the increasing number of patients, insufficient human or financial resources to provide the best quality services in a very short time frame.In order to implement and effectively use a web platform for screening, assessment and therapy of child and adolescent anxiety disorders, weran a survey among psychiatrists.We aimed for an overview on current practices in anxiety disorders (diagnosis, therapy, case management) of pediatric psychiatry specialized services across the country.Through the survey, we tried on one hand to evaluate the existing intervention in current practice used by child psychiatrists, and on the other hand, to assess their perception and willingness to use online assessment and intervention tool.

online platforme, assessment, intervention, child psychiatry

30. MANAGEMENTUL PACIENTILOR PEDIATRICI: TEHNOLOGIILE GENETICE SI GENOMICE CA INSTRUMENT DIAGNOSTIC

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Bolile de dezvoltare si malformatiile congenitale sunt printre cele mai comune afectiuni cronice pediatrice, cu efect sever atat asupra calitatii vietii pacientului, cat si a familiei sale. O provocare majora intalnita in practica si in cercetarea medicala este identificarea cauzelor acestor boli; factorii genetici si de mediu pot influenta multiple aspecte ale dezvoltarii organismului, respectiv ale formarii tiparelor comportamentale. Identificarea modificarilor genetice cauzative pentru aceste boli este instrumentala pentru diagnosticul si prognosticul pacientului, ca si pentru consilierea genetica a familiei. Ghidurile de buna practica si recomandarile curente subliniaza importanta diagnosticului precoce in optimizarea managementului clinic si a calitatii ingrijirilor medicale.

Peste 50 de pacienti cu diferite boli de dezvoltare si malformatii congenitale, au fost investigati prin cariotipare clasica, hibridizare fluorescenta in situ (FISH) si hibridizare comparativa genomica bazata pe microarray (array CGH, cariotip molecular pe platforma Agilent Technologies).

Au fost descrise anomalii genetice cauzative pentru sindroame precum: Williams, Angelman, Prader-Willi, DiGeorge, Miller-Diecker, Mowat-Wilson, de deletie 15q26-qter, de duplicatie a genei MECP2, de deletie 18q etc, ca si alte aberatii asociate cu dizabilitatea mentala idiopatica. Impactul diagnosticului genetic a fost mai mare in cazul pacientilor de varsta mai mica, unde abordarile terapeutice propuse au condus la imbunatatiri substantiale.

In concluzie, diagnosticul genetic este un instrument aflat la dispozitia clinicienilor din diverse discipline medicale in centrul carora se afla pacientul pediatric cu tulburari de dezvoltare, eficient pentru optimizarea managementului clinic si imbunatatirea calitatii vietii pacientului.
Suport financiar: PN 09.33.02.03
Cuvinte cheie: diagnostic genetic, FISH, cariotip molecular, boli de dezvoltare, management clinic

THE MANAGEMENT OF PEDIATRIC PATIENTS: GENETIC AND GENOMIC TECHNOLOGIES AS A DIAGNOSTIC TOOL

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Developmental disorders and congenital malformations are life-long impairing conditions, with high impact on the quality of life of both the patient and patient's family. A major challenge in the clinical practice is to identify the underlying causes; genetic and environmental factors may contribute in various degrees on a person's development and behavior. The identification of the causative genetic abnormality is instrumental for diagnosis, prognostic and for genetic counseling of the patient's family. Current clinical practice guidelines and recommendations emphasize the importance of early diagnosis in order to optimize the clinical management and patient care.
Over 50 pediatric patients with developmental disorders and congenital malformations were investigated by classical karyotyping, fluorescence in situ hybridization (FISH) and array-based comparative genomic hybridization (aCGH, Agilent Technologies).
Genetic defects causative for syndromes such as Williams, Angelman, Prader-Willi, DiGeorge, Miller-Diecker, Mowat-Wilson, 15q26-qter deletion, MECP2 duplication, 18q deletion etc, as well as other genetic aberrations associated with idiopathic intellectual disability were identified. The impact of the genetic diagnosis was highest for the youngest patients, where early therapeutic approaches resulted in substantial improvement.
Conclusions: genetic diagnostic is a powerful tool available to clinicians in many fields focusing on pediatric disorders, to increase the efficiency of patient's management and quality of life.
Acknowledgment: This work was supported by PN 09.33.02.03
Key words: genetic diagnostic, FISH, aCGH, developmental disorders, clinical management

31. SINDROMUL OPSOCLONUS-MIOCLONUS. CUM RECUNOAŞTEM? CUM EVALUĂM? CUM TRATĂM?

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Copil mic cu ataxie debutată recent, care poate duce la imposibilitatea mersului independent.
Ce este de făcut? Diagnosticul diferențial este vast: cele mai frecvente cauze sunt cele toxice, inflamatorii, vasculare, tumorale, dar nu trebuie să uităm de sindromul opsoclonus-mioclonus (SOM).
SOM este o boală mediată imun, fiind în multe dintre cazuri un sindrom paraneoplazic asociat unui neuroblastom. Este o afecțiune rară. Prognosticul său depinde de recunoașterea sa cât mai rapidă. Acest lucru este posibil dacă evaluarea clinică a copilului este completă și atentă la detaliile privind comportamentul motor, mișcările oculare, iritabilitatea și tulburările de somn. Atunci când există suspiciunea de SOM investigarea imagistică este foarte importantă și trebuie să fie țintită, pentru a confirma prezența unui neuroblastom, mai
frecvent abdominal sau toracal. Tratamentul este imunomodulator și chirurgical atunci când este cazul, cu o supraveghere clinică atentă, pentru a stabili pașii de urmat, în funcție de evoluția simptomatologiei.
Prin lucrarea de față dorim să prezentăm experiența clinicii noastre privind diagnosticul de SOM și modul în care evaluăm copiii clinic, investigațiile complementare pe care le propunem și tratamentul pe care îl inițiem, conform recomandărilor europene actuale.

32. FACTORI FAMILIALI SI PARENTALI IMPLICATI IN ETIOPATOGENIA ANOREXIEI NERVOASE LA ADOLESCENTI

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Lucrarea de fata isi propune sa identifice facorii de natura familiala si parentala intalniti in practica clinica la adolescentii diagnosticati cu anorexie nervoasa, cu accent pe interventia terapeutica in cadrul echipei multidisciplinare

formate din medic pediatru, medic pedopsihiatru şi psiholog. Dinamica si modelele de interactiune in familie s-au dovedit ca fiind cei mai importanti factori implicati etiologia tulburarilor de alimentatie in adolescenta.
Numeroase studii au confirmat astfel eficienta terapiei de tip sistemic si familial in tratamentul anorexiei nervoase a adolescentilor diagnosticati cu aceasta tulburare. Au fost descrise 3 tipologii familiale întâlnite la adolescenții diagnosticați cu anorexie, și anume: familia perfectă, cu expectanțe crescute din partea părinților adolescentul fiind cu o pesonalitate puternică si tendinte spre perfectiune; familia haotică, instabilă, abuzivă, adolescentul cu o personalitate ordonată, impulsivă; familia hiperprotectivă, simbiotică, ce neagă nevoile pacientului cu anorexie, cu o mamă hipercritică, eventual anorectică.
Vor fi analizate cateva cazuri clinice in care s-au identificat factorii parentali si familiali implicati in aparitia simptomatologiei si liniile de interventie terapeutica din perspectiva terapiei familiale sistemice.
Cuvinte cheie: anorexia nervoasa, adolescent, familie, terapie familiala

BOLI DE NUTRITIE SI METABOLISM / NUTRITION AND METABOLIC DISEASES

33. ROLUL POLIMORFISMELOR IL-6 572 C/G, 190 C/T, AND 174 G/C IN TULBURĂRILE DE NUTRIȚIE ALE COPILULUI

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Introducere Tulburarile nutritionale sunt afectiuni plurifactoriale in care aspectele genetice joaca un rol important alaturi de obiceiurile alimentare si stilul de viata. Inflamatia sistemica si nivelurile unor citokine sunt studiate in etiopatogenia acestor afectiuni. Am evaluat relația dintre polimorfismele genelor IL6-174G/C, IL6-190T/C si IL6-572G/C si starea de nutritie la un copil.
Metode Studiul a inclus 385 pacienti: malnutriti (173) , obezi (102) si martori (110), copii evaluati genetic, biochimic si antropometric.
Rezultate Pentru gena IL6-174G/C, la obezi a predominat genotipul CG (p=0,001); genotipul CC fiind factor de protecție, la malnutriti, genotipurile GG și CG (p=0,0001). Privind gena IL6-190T/C, genotipul CC a fost frecvent la obezi (p=0,0001), iar la malnutriti genotipul CT (p=0,003), genotipul TT avand functie protectiva. Analizand gena IL6-572G/C, genotipul CC fost mai frecvent la obezi (p=0,0001), GG predominand atat la malnutriti, cat si la martori. IMC, MUAC, TST și nivelurile albuminei s-au corelat cu genotipul CC al genelor IL6-190 și IL6-572 la obezi, iar la malnutriti, cu genotipurile GG si CG ale genelor IL6-174 si IL6-572 si cu genotipul CT al genei IL6-190.
Concluzii Tulburarile nutritionale la copii au fost mai frecvent asociate cu starea de purtator a alelei G IL6-174. Cel mai mare risc de a dezvolta obezitate a fost la purtatorii alelei C a genei IL6-572; genotipul CC al genei IL6-174 poate fi un factor protector fata de obezitate, iar genotipul IL6-190 TT poate avea functie protectoare pentru malnutritie. Cercetările ulterioare ar putea stabili rolul anumitor constelații genetice in tulburarile nutritionale.
Cuvinte cheie: copil, polimorfismul genic, tulburari nutritionale

THE ROLE OF IL-6 174 G/C, IL-6 190 G/C, IL-6 572 G/C GENES POLYMORPHISMS IN NUTRITIONAL DISORDERS OF CHILDREN

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Background Nutritional disorders are plurifactorial diseases in which the genetic aspects have an important role near the lifestyle and dietary habits. The systemic inflammation and cytokines levels are much studied in the etipatogenity of these diseases. We aimed to assess the relation between IL6-174G/C,IL6-190T/C andIL6-572G/C genes polimorphisms and nutritional status in a group of pediatric patients.
Methods The study included 385 patients: 173 malnourished, 102 obese and 110 controls in terms of genetic, biochemical and antropometric tests.
Results For *IL6-174G/C*gene, in obese, predominated of CG genotype (p=0.001); CC genotype was a protective factor, in malnourished, GG and CG genotypes (p=0.0001). Regarding *IL6-190T/C* gene, genotype CC was more frequent in obese (p=0.0001), in malnourished, CT heterozygotes (p=0.003), and TT genotype having protective function. Analysing *IL6-572G/C*gene polimorphisms, CC genotype was more frequent in obese (p=0.0001), GG predominated in both malnourished and controls. BMI, MUAC, TST and albumine levels correlated with CC genotype of IL6-190 and IL6-572 genes in obese,

while in malnourished, they correlated with GG and CG genotypes of IL6-174 and IL6-572 genes and with CT genotype of IL6-190 gene.
Conclusion: Child nutritional disorders were more frequent associated with IL6-174 G allele carriers. The highest risk of developing obesity was found in C-carriers of IL6-572 gene; CC genotype of IL6-174 may be a protective factor for obesity, while IL6-190 TT genotype could have protective function for malnutrition. Further research would establish the role of certain gene constellations in nutritional status disorders.
Keywords: child, gene polymorphism, nutritional disorders.

REUMATOLOGIE / RHEUMATOLOGY

34. REZULTATELE IMPLIMENTARII REGISTRULUI NAȚIONAL AL MALADIILOR REUMATICE LA COPII

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Introducere. Registrul national al maladiilor reumatice a fost implementat cu scopul documentarii bolilor reumatice pentru copii in Republica Moldova in anii 2012-2014.
Materiale si metode. In perioada anilor 2012-2014 grupul de lucru a colectat datele de baza clinice, paraclinice in urma aplicarii chestionarelor standartizate Childhood Health Assessment Questionnaire si Pediatric Quality of Life Inventory la 338 copii cu maladii reumatice.
Rezultate. La sfirsitul anului 2014 au fost inregistrati 338 copii cu maladii reumatice dintre care: 212 copii (63%) cu artropatii reactive, 91 copii (27%) cu artrita idiopatica juvenila, 34 copii (10%) cu maladii difuze ale tesutului conjuctiv. Repartitia genderica: 204 fetite (60%) si 134 (40%) – baietei. Durata medie de la debutul bolii pina la adresarea primara la specialist fost 11 luni. Durata medie a bolii a fost 25 luni. Afectarea articulara cu artrita activa a fost notata la 89% copii, iar cu deficit functional la 43% copii. Factorul rheumatoid pozitiv s-a determinat la 14,5% copii. Tratament de fond regulat au primit 37,3% copii.
Concluzii. Registrul bolilor reumatice la copii va facilita studiile clinice in domeniul reumatologiei pediatrice si va eficientiza managmentul copiilor cu maladii reumatice.

RESULTS OF IMPLEMENTING THE NATIONAL REGISTER OF RHEUMATIC DISEASES OF CHILDREN

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Introduction. The National Register of Rheumatic Diseases was implemented in order to document the rheumatic diseases of children in Moldova during the years 2012-2014.
Material and methods. During the period 2012-2014 the working group has collected the primary clinical and laboratory data through application of the standardized questionnaires Childhood Health Assessment Questionnaire and Pediatric Quality of Life Inventory on 338 children with rheumatic diseases.
Results. At the end of 2014 there were registered 338 children with rheumatic diseases including 212 children (63%) with reactive arthropathies, 91 children (27%) with juvenile idiopathic arthritis and 34 children (10%) with diffuse diseases of the connective tissue. Gender distribution: 204 girls (60%) and 134 (40%) boys. The average duration from the onset of the illness to the initial addressing to the doctor was 11 months. The mean duration of disease was 25 months. Articular damage with active arthritis was marked at 89% of children and functional deficit at 43% of children. The positive rheumatoid factor was positively determined at 14.5% of children. 37.3% of children received regular substance treatment.
Conclusions. The Register of Rheumatic Diseases of children will facilitate clinical studies in the field of pediatric rheumatology and will increase the efficiency in the management of rheumatic diseases of children.

35.IMUNODEFICIENTELE PRIMARE ASOCIATE BOLILOR AUTOIMUNE REUMATOLOGICE CU DEBUT JUVENIL: STUDIU MULTICENTRIC

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Introducere: Obiectivul nostru a fost acela de a investiga prevalenta si semnificatia clinica a imunodeficientelor primare (IDP) asociate bolilor autoimune reumatologice cu debut juvenil intr-un lot reprezentativ de pacienti.
Material si metode: Au fost inclusi in studiu 117 pacienti selectati consecutiv din 2 centre medicale tertiare pe parcursul unui an. Screeningul genetic al deletiei genei C2 de tip I si alotiparea genei C4 au fost efectuate prospectiv in toate cazurile. De asemenea, au fost notate retrospectiv nivelele serice ale imunglobulinelor investigate sistematic de-alungul evolutiei bolii.
Rezultate: Lotul nostru de pacienti cuprinde 84 cu artrita idiopatica juvenila (AIJ), 21 cu lupus eritematos sistemic (LES), 6 cu vasculita sistemica, 2 cu sclerodermie, 2 cu uveita idiopatica, 1 cu boala mixta de tesut conjunctiv si 1 cu overlap LES/sclerodermie. Au fost depistati 16 (13.7%) pacienti cu IDP, respectiv 7 cu deficit de C4, 5 cu deficit selectiv in IgA, 3 cu deficit in C2 si 2 cu hipogamaglobulinemie neincadrata (unul a asociat si deficit de C4). In cadrul celor 84 pacienti cu AIJ, 4.8% au avut deficite congenitale de complement, prevalenta mult inferioara celei din cadrul cohortei cu LES (23.8%), dar toti au demonstrat o evolutie agresiva a bolii. Majoritatea pacientilor cu IDP ale anticorpilor au avut o evolutie severa grefata de complicatii si chiar asociere de 2 patologii autoimune.
Concluzie: Rezultatele studiului noastru sustin necesitatea de a evalua, in cazul pacientilor cu boli autoimune reumatologice, posibilele IDP asociate, avand in vedere prevalenta si aparenta lor implicare in formele severe de boala.

PRIMARY IMMUNODEFICIENCIES IN AUTOIMMUNE RHEUMATOLOGIC DISEASES WITH JUVENILE ONSET: MULTICENTER STUDY

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Background: Our aim was to investigate the prevalence and clinical relevance of inherited complement and antibody deficiency states in a large series of patients with various autoimmune rheumatologic diseases (ARD) with juvenile onset.
Material and methods: A total number of 117 consecutive patients from 2 tertiary referral hospitals were included in the study. All patients underwent genetic screening for type I C2 deficiency and C4 allotyping. Serum levels of immunoglobulin classes measured systematically throughout their regular medical care were recorded retrospectively.
Results: Our cohort of patients included 84 with juvenile idiopathic arthritis (JIA), 21 with systemic lupus erythematosus (SLE), 6 with systemic vasculitis, 2 with juvenile scleroderma, 2 with idiopathic uveitis, 1 with mixed connective tissue disease and 1 with SLE/scleroderma overlap syndrome. We have found 16 patients with evidence of primary immunodeficiency in our series (13.7%), including 7 with C4 deficiency, 5 with selective IgA deficiency, 3 with C2 deficiency and 2 with unclassified hypogammaglobulinemia (one also presented C4D). Of the 84 patients with JIA, 4.8 % had complement deficiencies, which was less prevalent than in the SLE cohort (23.8%), but all of them have exhibit an aggressive disease. Most of our patients with primary antibody deficiencies showed a more complicated and severe disease course and even the co-occurrence of two associated autoimmune diseases.
Conclusions: Our findings among others demonstrate that complement and immunoglobulin immunodeficiencies need careful consideration in patients with ARD, as they are common and might contribute to a more severe clinical course of the disease.

36. REACTIE ADVERSA LA METHOTREXAT-TOXICITATE HEMATOLOGICA

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Autorii prezinta, cazul unei adolescente ,aflate in evidenta speciala a sectiei Clinicii Pediatrie din cadrul SCJU Tg.Mures cu diagnosticul Artrita juvenila idiopatica forma poliarticulara seropozitiva, in tratament cronic cu Methotrexat de aproximativ doi ani. In urma reevaluarilor clinico-biologice periodice, s-au constatat semne si simptome clinice confirmate paraclinic, datorate reactiei adverse la Methotrexat , manifestate prin toxicitate hematologica.

ADVERSE REACTION TO METHOTREXATE - HAEMATOLOGICAL TOXICITY (CASE REPORT)

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The authors present the case of a teenager in the records of the Pediatric Clinic no. I Tg-Mureş, diagnosed with seropositive polyarticular juvenile idiopathic arthritis treated with methotrexate for about two years. Following clinical and biological periodic revaluations were found signs and clinical symptoms of haematological toxicity of methotraxat considered adverse reaction to the medication, confirmed with laboratory evaluations.

URGENŢE – TOXICOLOGIE / PEDIATRIC EMERGENCIAS – CLINIC TOXICOLOGY

37. SERVICIUL DE SFAT MEDICAL TELEFONIC IN URGENTA LA SPITALUL CLINIC DE COPII CLUJ – RISCURI VERSUS BENEFICII

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In lipsa unui triaj eficient la nivelul retelei de medicină primara, supra-aglomerarea UPU si a camerelor de garda de pediatrie a devenit o realitate incontestabila, multe dintre prezentări nefiind cu adevarat urgente din punct de vedere medical.
Autorii prezinta rezultatele obtinute după 12 luni de activitate a serviciului de sfat medical telefonic care funcționează in cadrul Spitalului Clinic de Urgenta pentru Copii Cluj-Napoca. Acest serviciu a functionat cu finantare privata si cu acordul Ministerului Sanatatii, incercand sa trieze cazurile care necesitau consult medical imediat, in primele 2 ore, respectiv in primele 24 ore de la apel. Un numar de 35 medici cu specialitatea Pediatrie sau Medicina de Urgența au raspuns unui numar de 8818 apeluri provenite in cea mai mare parte (peste 95%) din județul Cluj. Principalele cauze de apelare au fost (in ordine): febra, varsaturile, tusea, diareea, eruptiile cutanate, 68% din totalul apelurilor finalizandu-se cu recomandari de tratament la domiciliu si informarea apartinatorilor asupra semnelor de alarma.
Dupa un an de activitate, s-a observat: scaderea numarului de cazuri prezentate la camerele de garda si reducerea numarului de ambulante trimise de SAJ Cluj la cazurile pediatrice. Nu s-au inregistrat reclamatii referitoare la aspectele medicale ale sfatului telefonic, iar rata de satisfactie a apelantilor a fost de peste 97%. In concluzie, serviciul de sfat medical telefonic reprezinta un sistem eficient de triaj telefonic al afectiunilor acute pediatrice, aspect care a fost semnalat si Ministerului Sănătății in ideea preluarii finanțarii necesare pentru continuarea activității.

TELEPHONE MEDICAL ADVICE SERVICES IN EMERGENCIES AT CLUJ PAEDIATRIC HOSPITAL - RISKS VERSUS ADVANTAGES

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Due to a lack of an efficient selection at the level of primary medicine network, the Emergency Unit and the paediatric examination rooms' on-duty overcrowding, became an unquestionable reality, many of the cases not being a real emergency from medical point of view.
The authors reveal the results obtained after 12 months of activity of telephone medical advice services running within Cluj-Napoca Children's Emergency Clinical Hospital. The service ran its activity with private financing and with Ministry of Health's approval, trying to select the cases needing immediate medical examination, in the first 2 hours and the first 24 hours respectively from the call. A number of 35 physicians having Paediatrics or Emergency Medicine as specialties attended a number of 8,818 calls coming mostly (over 95%) from Cluj County. The main reasons for calling were (in order): fever, vomiting, coughing, diarrhoea, coetaneous eruptions, 68% of the total calls ending with home treatment recommendations and informing the callers on the alarming signs.
After a year of activity we noted: the number of cases for the examination rooms on-duty diminished and also the number of ambulances dispatched by SAJ Cluj for paediatric cases. Any complaints were recorded concerning the medical aspects of telephone advice service and the rate of callers' satisfaction was more than 97%. In conclusion, the telephone medical advice is an efficient system of paediatric acute disorders telephone selection, issue of which the Ministry of Health was informed of, in order to take over the needed financing for the service to go on.

38. INTOXICAŢIILE ACUTE CU MEDICAMENTE CARDIOTOXICE. STUDIU MULTICENTRIC RETROSPECTIV – BUCUREŞTI, CLUJ-NAPOCA, IAŞI SI TIMIŞOARA.

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8. Universitatea de Medicina si Farmacie "Carol Davila" Bucuresti

Introducere. Numeroase medicamente aparţinând claselor farmacologice cardiovasculare şi noncardiovasculare pot afecta în doze toxice buna funcţionare a aparatului cardiovascular. Inima şi muşchiul neted vascular pot fi agresate de toxic în mod direct, dar şi indirect prin interferarea cu procesele de reglare simpatică şi parasimpatică sau prin alterarea volumului intravascular şi a echilibrului metabolic. Material şi metodă. În studiul de faţă autorii şi-au propus aprecierea caracteristicilor demografice, etiologice şi circumstanţiale ale intoxicaţiilor acute cu medicamente cardiotoxice, precum şi caracterizarea

profilului clinic şi electrocardiografic al acestor pacienţi. Pentru realizarea acestor obiective am adunat retrospectiv datele din 4 centre antitoxice de referinţă - Bucureşti, Cluj-Napoca, Iaşi si Timişoara – pe o perioadă de 3 ani (2012-2014). Rezultate. În perioada studiată au fost internaţi 443 copii şi adolescenţi cu intoxicaţie acută cu medicamente cardiotoxice. Au predominat intoxicatiile acute cu anticonvulsivante (19%) si ingestiile plurimedicamentoase (19%). Am observat 2 vârfuri de incidenţă în grupa de vârstă 1-5 ani şi, respectiv 11-18 ani, precum şi o frecvenţă mai mare a sexului feminin (63%). 56% din cazuri au fost expuneri voluntare. Principala manifestare clinică observată a fost tahicardia (58%), urmată de bradicardie (27%) şi hipotensiune arterială (22%). Principalele modificări electrice înregistrate au fost tulburările de conducere intraventriculară (12%) şi alungirea intervalului QTc (9%). Şocul cardiogenic şi/sau vasoplegic a fost prezent în 8 cazuri. Concluzii. Afectarea funcţiei cardiovasculare la copiii şi adolescenţii cu intoxicaţie acută reprezintă o complicaţie gravă, ameninţătoare de viaţă. Abordarea diagnostică şi terapeutică a acestor cazuri trebuie să fie promptă, adesea într-o unitate de terapie intensivă.

ACUTE POISONING WITH CARDIOTOXIC DRUGS. A RETROSPECTIVE MULTICENTRE STUDY - BUCURESTI, CLUJ-NAPOCA, IASI AND TIMISOARA.

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Background. Many drugs from cardiovascular and non-cardiovascular pharmacologic classes can affect in toxic doses the proper functioning of the cardiovascular system. The heart and the vascular smooth muscle can be aggressed by the toxic both directly and indirectly by interfering with sympathetic and parasympathetic regulation processes or by altering intravascular volume and metabolic balance. Material and methods. In this study the authors have planned to assess demographic, etiologic and circumstantial features of acute poisoning with cardiotoxic drugs and to characterize the clinical and electrocardiographic profile of these patients. In order to achieve these objectives we collected data retrospectively in 4 antitoxic reference centres - Bucharest, Cluj-Napoca, Iasi and Timisoara - for a period of three years (2012-2014). Results. During the study 443 children and adolescents were hospitalized for acute poisoning with cardiotoxic drugs. Acute poisoning with anticonvulsants (19%) and multidrug ingestion (19%) predominated. We observed two peaks of incidence in the age group 1-5 years and 11-18 years respectively, and a higher frequency of females (63%). 56% of cases were intentional exposures. The main clinical sign observed was tachycardia (58%), followed by bradycardia (27%) and hypotension (22%). The main electrical changes recorded were intraventricular conduction disturbances (12%) and QTc prolongation (9%). Cardiogenic and/or vasoplegic shock was present in 8 cases. Conclusion. Impairment of cardiovascular function in children and adolescents with acute poisoning is a serious, life-threatening complication. Diagnostic and therapeutic approach to these cases should be promptly, often in an intensive care unit.

39. BAUTURILE ENERGIZANTE CHIAR NU SUNT NOCIVE ?

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Desi nu exista consens asupra definitiei bauturilor energizante, se accepta ca acestea sunt bauturi care contin cafeina, taurina , una sau mai multe vitamine si in unele cazuri o sursa de energie (ex. glucide) si/sau una sau mai multe alte substante comercializate in scopul declarativ de crestere a performantelor fizice si intelectuale. De la prima bautura energizanta comercializata pentru prima data in Austria in 1987 , s-a ajuns in prezent la citeva sute de preparate. Copiii si adolescentii de sex masculin sunt cei mai mari adepti al acestor bauturi. Interesul toxicitatii este axat pe principalul ingredient al lor si anume cafeina , al carui consum maxim la copil si adolescent nu trebuie sa depaseasca zilnic 2,5 mg /kg. Studiile arata ca toxicitatea se manifesta prin simptome nervoase
(somnolenta, vertij, acufene, tulburari vizuale, confuzie , agitatie), cardiovasculare (HTA, aritmii ventriculare sau supraventriculare, ischemie miocardica) digestive (varsaturi, diaree, dureri abdominale), rabdomioliza cu edem pulmonar, insuficienta renala si chiar deces.
In prezent cel mai in voga cocktail in rindul tinerilor este amestecul bauturilor energizante cu votca sau whiski . Amestecul de alcool cu bauturi energizante poate masca semnele unei intoxicatii etanolice acute, prin intirzierea aparitiei efectelor depresoare asupra sistemului nervos central, ceea ce faciliteaza consumul unei cantitati si mai mari de alcool cu toate consecintele sale. Un alt obicei este asocierea lor cu amfetamine, in acest caz existind risc de convulsii si hipertermie maligna.
Cuvinte cheie : energizante, cafeina, toxicitate

ENERGY DRINKS, REALLY HARMFUL ?

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Although there is no consensus on the definition of energy drinks, it is accepted that these are drinks that contain caffeine, taurine, one or more vitamins and in some cases an energy source

(ex. Carbohydrates) and / or one or more other substances marketed declaratory order to increase mental and physical performance. Since the first commercialized energy drink in Austria in 1987, it has now reached several hundred preparations. Children and young male are the largest followers of these beverages. Toxicity is focused on their main ingredient, namely caffeine, whose maximum consumption in children and adolescents should not exceed the daily 2.5 mg / kg. Studies show that toxicity is manifested by symptoms of nervous system (Somnolence, vertigo, tinnitus, visual disturbances, confusion, agitation), cardiovascular system (hypertension, supraventricular or ventricular arrhythmias, myocardial ischemia) gastrointestinal system (vomiting, diarrhea, abdominal pain), rhabdomyolysis with pulmonary edema, kidney failure and even death.
Currently the hottest cocktail among young people is mixing energy drinks with vodka or whiskey. The mixture of alcohol with energy drinks can mask signs of acute ethanol poisoning, will delay depressant effect of the central nervous system, which facilitates greater quantities and consumption of alcohol with all its consequences. Another custom is their association with amphetamines, in this case the risk of malignant hyperthermia and convulsions.
Keywords: energy, caffeine, toxicity

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40. COMELE TOXICE LA COPII – STUDIU PE O PERIOADA DE 9 ANI

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Coma este manifestarea clinica a unui larg spectru etiologic, iar etiologia toxica reprezinta cauza principala a comei la copii.
Scopul si obiectivele: Studiul efectuat, de tip retrospectiv, a urmarit toate cazurile de come prezentate in Unitatea de Primire Urgente a Spitalului “Grigore Alexandrescu” Bucuresti, pe o perioada de 9 ani, din 2003 pana in 2011. Din totalul comelor le-am selectat pe cele de origine toxica si le-am analizat din punct de vedere al etiologiei, profunzimii, tipului si al repartitiei în functie de varsta, sex, intentie. Scopul studiului a fost identificarea si diagnosticarea rapida din punct de vedere etiologic a comelor toxice la copii si a factorilor de risc asociati, in vederea instituirii unui tratament precoce si eficient. De asemenea am analizat factorii de risc asociati comelor toxice care evolueaza catre deces, precum si semnele clinice care insotesc alterarea starii de constienta, la prezentarea in camera de garda. Am studiat si relatiile intre etiologie si timpul scurs pana la revenirea starii de constienta si timpul de la instalarea comei pana la prezentarea in UPU.
Concluzii: In ultimii ani se constata o crestere a frecventei intoxicatiilor acute la copil, determinate in principal de medicamente si substante toxice de uz casnic, dar si de alcool si droguri. Pe langa faptul că a crescut numarul total de intoxicatii la copii, si gravitatea lor a crescut si unul dintre elementele care sustine acest lucru este reprezentat de frecventa in crestere a comelor toxice.
Cuvinte cheie: coma, toxic, diagnostic etiologic, tratament

TOXIC COMA IN CHILDREN – A 9 YEARS RETROSPECTIVE STUDY

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Coma is a clinical manifestation of a large etiological spectrum, and toxic etiology is the leading cause of coma in children.
Methods: In our retrospective study, we registred all children with coma examined in the Emergency Department of “Grigore Alexandrescu” Children Hospital, during a nine year period, from 2003 to 2011. We focused on the toxic coma and we analised data using the following criteria: etiology, depth, distribution by age, gender, reason of presentation. The aim of study was to identify the etiology of toxic coma and related risk factors, for establish an early and effective treatment. We also analised the risk factors associated to toxic coma evolving to death and also the clinical feature that accompany altered consciousness at the admitted time. We studied the connection between etiology and return to consciounsness and the time since start of coma and the moment of presenting in emergency room.
Conclusion: In the last few years, we saw a frequency increase of acute poisoning in children, caused mainly by drugs, toxic household substances, alcohol and abuse substances. Not only the total number of acute poisoning is incresed, but their severity is also increased and toxic coma represents a strong example.
Key words: coma, toxic, etiology, treatment

41. PROFILUL BIO-PSIHO-SOCIAL AL TINERILOR CONSUMATORI DE SUBSTANTE PSIHOACTIVE

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Scop
Determinarea ponderii consumatorilor de substanţe psihoactive (SP). Evaluarea factorilor de risc pentru adolescenţii consumatori de SP.
Material si metode
Populaţia ţintă este reprezentată de totalul de internărilor pe o perioadă de 5 ani (1 ianuarie 2009 – 31 decembrie 2013) cu diagnosticul de intoxicaţie voluntară. Studiul a extras un lot de 42 de pacienți internați în Spitalul de Copii „Louis Țurcanu” Timișoara. Variabilele au fost: vârsta adolescenţiilor, sexul, mediul de provenienţă, tipul de familie din care provin, nivelul de inteligenţă, de școlarizare, diagnosticul psihiatric.
Rezultate si discutii
Frecvenţa dominantă este reprezentată de intoxicaţiile cu medicamente, cu un procent de 45,6%. Etnobotanicele, cea mai frecventă substanţă psihoactivă utilizată de pacienții cuprinși în studiu**.** Majoritatea pacienţilor asupra cărora s-a efectuat acest studiu au o vârstă cuprinsă între 14 si 18 ani si provin preponderent din mediul urban. Sub 10% dintre pacienţi sunt cuprinsi in sistemul de educaţie, 88% din ei fac parte dintr–o familie dezorganizată.
Concluzii
Consumul de SP este un real pericol prin amploarea pe care a dobândit-o între tinerii români, fiind și în crestere. Rata cea mai mare de creştere a incidenţei intre 2009- 2012 apartine etnobotanicelor. Adolescenții dependenți de droguri au o rată crescută de abandon școlar.
Cuvinte cheie - substante psihoactive, tineri, intoxicatii

BIO-PSYCHO-SOCIAL PROFILE OF YOUNGSTERS USERS OF PSYCHOACTIVE SUBSTANCES

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The aim was estimation of the share of consumers of psychoactive substances (SP) and assessment of risk factors for adolescent consumers of SP.
Material and Methods
The target population is the total number of admissions over a period of 5 years (1 January 2009 - 31 December 2013) with the diagnosis of voluntary intoxication. The study extracted a sample of 42 patients admitted to Children's Hospital "Louis Turcanu" Timișoara. The variables were: teenagers age, sex, origin, type of family they come from, level of intelligence, education, psychiatric diagnosis.
Results and discussion
Dominant frequency from all was the drugs poisoning, with a percentage of 45.6%. Etnobotanical were the most commonly used psychoactive substance in the study patients. Most patients on this study which was carried out between the ages of 14 and 18 and come mainly from urban areas. Less than 10% of patients are enrolled in the education system, 88% of them are part of a broken home.
Conclusion
SP consumption is a real danger by the extent between Romanian teenagers, and also the rise. The highest rate of increase in incidence between 2009- 2012 was SP poisoning. Teenagers addicted to drugs have a high rate of school dropout.
Key words - PSYCHOACTIVE, youngsters, poisoning

POSTERE PREZENTATE ORAL / POSTERS SELECTED FOR ORAL PRESENTATION

ALERGOLOGIE / ALLERGOLOGY

1. UTILITATEA PANELURILOR ALERGOLOGICE IN PATOLOGIA ATOPICA PEDIATRICA – STUDIU RETROSPECTIV

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Introducere. Profilul alergologic poate fi studiat folosind investigatii de tip *panel* care sunt mai accesibile decat testele alergologice cutanate. Obiective. Evaluarea utilitatii panelurilor pediatrice in managementul copiilor cu manifestări alergice internati intr-o Clinica de Pediatrie. Material si metoda. Au fost studiate retrospectiv panelurile utilizate pe parcursul a 12 luni la copiii cu patologie atopică (astm, dermatita, conjunctivita atopica, alergii alimentare). Evaluarea statistica a fost realizata cu MS Office si EpiInfo. Rezultate. 145 de copii (77 baieti) cu varsta de peste 4 ani au fost inclusi in studiu. Dintre copiii cu astm (101 cazuri) 44 au avut rezultate semnificative la panel vs. 10 din copiii non-astmatici: p = 0,008, OR = 2,62 (1,17-5,88). Sensibilizarea la acarieni a fost cel mai frecvent intalnita, urmata de cea la proteinele din lapte (beta si alfa-lactalbumina). In cazul alergiilor alimentare (12 cazuri) numai 2 cazuri au avut concordanta intre proba clinica si valoarea IgE specifice. Concluzii. Panelurile alergologice sunt modificate mai ales la copiii cu patologie astmatica. In cazul alergiilor alimentare proba clinica este mai importanta decat valorile IgE specifice.
Cuvinte cheie: copil, atopie, IgE specific

ALLERGY PANELS IN CHILDREN ADMITTED TO A PEDIATRIC CLINIC – RETROSPECTIVE STUDY

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Introduction. The atopic status may be evaluated using allergy panels which are more accessible than the classical skin tests. Objectives. To evaluate the utility of the pediatric panels for the management of the atopic children admitted to our clinic. Methods. The allergic patients (asthma, atopic rhinitis, atopic conjunctivitis and food allergies) admitted during one year were retrospectively studied. The statistical analysis was done using MS Office and EpiInfo. Results. 145 children (77 boys) aged over 4 completed the survey. The positive panel results emerged in 101 of the asthmatic children vs. 10 cases in the non-asthma group: p = 0.008, OR = 2.62 (1.17-5.88). Sensitization to dermatophagoides (both types), followed by the milk proteins were the most frequent encounters. Only two cases of food allergies showed concordance between the oral food challenge and the specific IgE results. Conclusions. The allergy panels were positive mostly in asthma cases. The food allergies were diagnosed using the clinical challenge rather the specific IgE levels.
Key words: child, atopy, specific IgE

CARDIOLOGIE / CARDIOLOGY

2. MALFORMATII ATRIALE STANGI COMPLICATE CU HIPERTENSIUNE PULMONARA DE TIP VENOS

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Spitalul de Copii"Louis Turcanu" Timisoara

Scop: Descrierea manifestarilor clinice, ecografice si evolutive in cazul unor malformatii rare ale atriului stang insotite de hipertensiune pulmonara de tip venos.
Material: Prezentam cazurile a 3 copii (raport sexe M:F = 1:2), care s-au prezentat cu hipertensiune pulmonara severa cu cianoza, polipnee, fatigabilitate marcata la supt. In cel de-al 3-lea caz o tuse iritativa persistenta de aproape 2 luni, cu moderata scadere a capacitatii de efort au fost semnele de debut.
Varsta de debut a fost 1; 3 luni respectiv 3 ani. Ecografic s-a evidentiat un drenaj venos total aberat de tip supracardiac, vena colectoare stenozata drena in vena cava superioara si un defect septal atrial cu shunt dreapta stanga.

Un al II-lea caz a fost diagnosticat un cor triatrial cu o membrana in atriul stang creeind gradient transmembranal de 16 mm Hg, cu dilatare si disfucntie de ventricul drept.
Cel de-al III-lea caz a fost depistat cu valva mitrala dubla si insuficienta mitrala.
Rezultate : In primul caz s-a reimplantat vena colectoare in atriul stang simultan cu inchiderea defectului septal atrial. Al II-lea copil a beneficiat de excizia membranei obstructive din atriul stang si sutura defectului atrial. In ambele cazuri evolutia postoperatorie a fost rapid favorabila cu reducere presiunilor pulmonare si dimensiunilor ventricolului drept. Ultimul caz a avut o evolutie buna sub tratament cu diuretic.
Concluzie: Diferite malformatii intersand atriul stang pot determina hipertensiune pulmonara de tip venos la copil. Diagnosticul precoce este esential, fiind salvator.

LEFT ATRIAL MALFORMATIONS WITH PULMONARY HYPERTENSION OF VENOUS TYPE

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Children's Hospital "Louis Turcanu" Timisoara

The aim of the study was the description of the clinical manifestation, echocardiography and follow-up in case of some rare left atrial malformation, accompanied by venous pulmonary hypertension.
Material and method: We present 3 children (male:female ratio1:2), 2 infants who presented with severe pulmonary hypertension, cyanosis, polypneea , fatigability at eating. The third case the presenting sign was a 2 month old persisting cough, exertion. Symptoms started at the age of 1, 3 month, 3 years respectively. A total abnormal venous drainage of supracardiac type (the collector draining in the superior vena cava with stenosis) and atrial septal defect was detected in the first case. A cor triatriatum with atrial septal defect was present in the second case. The transmembrane gradient was 16 mm Hg, with dilatation and dysfunction of the right ventricle. The third boy was diagnosed with a double mitral valve and grade II/III mitral insufficiency and pulmonary stasis.
Results: A implantation of the collector vein in the left atrium with closure of the atrial defect (ASD) was performed in the first infant. Membrane resection in cor triatriatum and closure of ASD was done in the second case. A rapid improvement with reduction of the pulmonary pressures and of the dimensions of the right ventricle followed. The third case only diuretic medication was helpful.
Conclusion: Different malformation of the left atrium may cause venous pulmonary hypertension in children. An early diagnosis is mandatory and life saving.

3. ASPECTE ALE AFECTĂRII CARDIACE LA COPIII CU SINDROM WILLIAMS-BEUREN

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Sindromul Williams-Beuren este o afectiune genetica relativ frecventa, determinata de microdeleţia 7q11.23 si manifestata clinic prin dismorfie craniofaciala caracteristica, malformatie cardiaca si dizabilitate intelectuala cu personalitate comunicativa. Uneori se poate asocia deficit de crestere, hipercalcemie sau anomalii de tesut conjunctiv. Obiective: Evaluarea modificarilor cardiace la un lot de copii diagnosticati cu sindrom Williams-Beuren la Cabinetul de Genetica Medicala a spitalului nostru. Material si metoda: Lotul de studiu a cuprins un număr de 31 copii internaţi într-o perioadă de 10 ani. Acestor pacienti li s-au efectuat: examen clinic (inclusiv masurarea TA), testări biologice (calcemie), ECG, ecocardiografie şi examen genetic. Confirmarea diagnosticului de sindrom Williams a fost efectuata prin testari moleculare FISH si MLPA. Rezultate: Ecocardiografia a relevat stenoza de aorta supravalvulara in 46.6% cazuri, coarctatie de aorta in 46,6% cazuri, prolaps de valvă mitrala in 33,3% cazuri si in proportie mai mica defect septal ventricular, persistenta de canal arterial, defect septal atrial, bicuspidie aortica, stenoza pulmonara si defecte combinate. O parte dintre copii au beneficiat de interventie chirurgicala corectoare. Concluzii: Pacientii care prezinta dismorfie faciala sugestiva, dizabilitate intelectuala medie si suflu sistolic trebuie investigati ecocardiografic si din punct de vedere genetic pentru diagnosticarea sindromului Williams, in scopul urmaririi ulterioare si evitarii complicatiilor posibile (afectare cardiaca severa, litiaza renala, hipotiroidie, hipertensiune, risc anestezic). Uneori tabloul clinic poate fi incomplet, dar testul genetic (facil, economic) poate duce la finalizarea diagnosticului, cu prevenirea complicatiilor.

CARDIAC INVOLVMENT IN CHILDREN WITH WILLIAMS-BEUREN SYNDROME

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Williams-Beuren syndrome is a relatively frequent genetic disorder caused by 7q11.23 microdeletion and characterized by facial dysmorphism, cardiac malformation and intelectual disability with communicative personality. Sometimes short stature, hypercalcemia and connective tissue abnormalities can be associated. Objective: assesment of cardiac anomalies in a group of patients diagnosed with Williams syndrome in the Medical Genetic Lab of our hospital. Method: the studied group

consisted in 31 patients addmited in a 10 years period. We performed clinical examination, including blood pressure measurements, biological tests (serum calcium), ECG, echocardiography and genetic exam. The genetic diagosis was established by Fluorescence-in-situ-hybridization (FISH) and Multiple Ligation Probe Amplification (MLPA). Results.echocardiography revealed supravalvulr aortic stenosisin 46.6% cases, aortic coarctation in a 46.6% cases, mitral valve prolapse in 33.3% cases and, in a smaller percent, ventricular septal defect, bicuspid aortic valve, pulmonary stensis and also combinations of the mentioned malformations. Some of the patients had corrective surgery. Conclusions. The patients with suggestive facial dysmorphism, medium intelectual disability and cardiac murmur must receive an echocardiography and genetic tests for Williams syndrome in order to follow-up and avoid future possible complications (severe cardiac suffering, hypertension, renal lithiasis, hypothyroidism, anesthesia risk). Sometimes the clinical picture can be incomplete, but the genetic test (easy to perform and cheap) can finalize the diagnosis and prevent the complications.

ENDOCRINOLOGIE ŞI DIABET PEDIATRIC / PEDIATRIC ENDOCRINOLOGY AND DIABETED

4. UN PROFIL GLICEMIC CIUDAT – INDICIU DIAGNOSTIC LA UN PACIENT NEDIABETIC

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Un sugar de sex masculin în vârstă de 21 de zile, alimentat mixt, s-a internat în Unitatea de Primire Urgențe pentru o curbă ponderală nesatisfacătoare și inapetență. Istoricul personal și familial nu au identificat elemente semnificative. La internare avea semne clinice de deshidratare, prezenta un icter moderat și o hepatomegalie marcată. Explorările de laborator au evidențiat un sindrom inflamator, hepatocitoliză și colestază, hiperbilirubimenie indirectă și un sindrom de retenție azotată. S-a stabilit diagnosticul de septicemie neonatală, hepatită colestatică și insuficiență renală acută. La transferul în clinica de pediatrie o glicemie determinată cu bandelata a arătat o valoare de 72 mg%. A primit perfuzii cu glucoză și electoliți, antibioticoterapie și gamaglobuline intravenos. S-au exclus infecțiile TORCH. Analiza LCR a fost negativă. După internare a prezentat și diaree. Nu a mai fost alăptat datorită hipogalactiei mamei și a primit o formulă de lapte fără lactoză. Sub tratament, apetitul se reia, crește în greutate și funcția hepatică se ameliorează. S-a externat pe propria răspundere, dar revine după o săptămână cu vărsături și deshidratare. Profilul glicemic efectuat cu bandeleta a arătat valori constant peste 200 mg%. Dozarea glicemiei serice a demonstrat de fapt glicemii de 60 mg%. În urină s-a demonstrat prezența de substanțe reducătoare și s-a exclus glicozuria. Actvitatea scăzută a galactozo 1 fosfat uridyltransferazei a stabilit diagnosticul de galactozemie. Rezultatele screeningului neonatal au fot disponbile deabia dupa 2 luni.Valorile discordante ale glicemiei determinate cu glucometrul față de glicemia determinată din ser au reprezentat un indiciu decisiv pentru diagnosticarea galactozemiei.
Cuvinte cheie: hiperglicemie, hipoglicemie , galactozemie

A STRANGE GLYCEMIC PROFILE IN A NON DIABETIC INFANT AS A DIAGNOSTIC CLUE

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A 21 day old boy, on combined breastfeeding and standard formula, was admitted in the emergency care unit for poor weight gain and lack of appetite. The pregnancy and family history were unremarkable. He presented clinical signs of dehydration, mild jaundice, marked hepatomegaly. The investigation showed: inflammatory syndrome, elevated transaminases, cholestasis with mild unconjugated hyperbilirubinemia, elevated urea and creatinine. The diagnostic of newborn septicemia, cholestatic hepatitis and acute renal failure was formulated. He was transferred in the Department of Pediatrics. A glucose meter check showed a value of 72 mg%. He received glucose and electrolyte perfusions, antibiotic therapy and intravenous immunoglobulin. We ruled out the TORCH infections. After admission, he associated loose stools. His breast feeding was discontinued due the hipogalactia and the standard formula was replaced with a lactose-free formula. The CSF analysis was negative. Under treatment, he had an improvement in feeding, growth and liver function tests. The patient was discharged against medical advice. After a week, he was readmitted for vomiting and dehydratation. Repeated glucose meter checks performed overnight were all greater than 200 mg/dl. A serum sample sent to the laboratory showed the glucose to be 60 mg%. The urine was strongly positive for reducing substances and negative for glucose. The red blood cell study of galactose-1-phosphate uridyl transferase (GALT) activity was confirmatory. The result of the neonatal screening confirming the galactosemia was available only 2 months after. The discordant glucose meter and serum sample glucose concentrations provided an important clue to the diagnosis of galactosemia. Keywords: hypoglicemia, Hyperglicemia, galactosemia

5. BOLI AUTOIMUNE ASOCIATE DIABETULUI DE TIP 1 LA COPIL

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Introducere. Bolile autoimune se asociaza frecvent diabetului de tip 1. Diagnosticate precoce, pot preveni aparitia complicatiilor, de aceea screeningul initial reprezinta o optiune importanta la copiii cu diabet zaharat la debut.
Obiective Ne-am propus sa evaluam prezenta autoimunitatii specifice si asocierea bolilor autoimune (tiroidita si celiachie) la copiii recent diagnosticati cu diabet de tip 1.
Material si metoda Studiul a fost efectuat in Clinica de Pediatrie din Constanta, in perioada ianuarie 2011- mai 2015, pe un lot de 43 de copii cu diabet de tip 1.
La debutul diabetului am evaluat urmatorii anticorpi: antiglutamat decarboxilaza (GAD-II), anti-tirozinfosfataza IA2, anti-insulina, anti-tireoperoxidaza, anti-tiroglobulina, antitransglutaminaza IgA si IgG.
Rezultate Prezenta autoanticorpilor specifici diabetului a fost evaluata la 20, din cei 43 de pacienti. 2 pacienti (10%) au avut 3 anticorpi pozitivi, 9 au avut 2 anticorpi pozitivi (45%) , 8 pacienti cu 1 anticorp pozitiv (40%) si 2 cu anticorpi negativi (10%). Din cele trei tipuri de anticorpi cel mai frecvent este antiglutamat decarboxilaza (GAD-II) 14 cazuri.
Screening-ul tiroidian:20.9% au prezentat cel putin unul dintre anticorpi pozitivi la de debutul diabetului. 1 pacient a dezvoltat ulterior disfunctie tiroidiana.
Boala celiaca a fost descoperita la 6.96% dintre subiecti. Unul a prezentat initial anticorpi antitransglutaminaza borderline, cu pozitivare la un an de la diagnosticarea diabetului. 2 dintre pacienti au efectuat endoscopie digestiva superioara si biopsie duodenala, constatandu-se histologic stadiul Marsh II.
Concluzii
Detectarea autoanticorpilor, inca de la debutul diabetului, previne instalarea complicatiilor determinate de intarzierea diagnosticarii bolilor asociate.
Cuvinte cheie:diabet, copil, celiachie, tiroidita, autoimunitate

AUTOIMMUNE DISEASES ASSOCIATED WITH TYPE 1 DIABETES IN CHILDREN

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Introduction
Autoimmune diseases are often associated with type 1 diabetes. Early diagnosis can prevent complications; therefore, initial screening is an important option for children at the onset of the disease.
Objectives. We proposed to evaluate the presence of specific autoimmunity and the association of autoimmune diseases (thyroiditis and Celiac disease) in children diagnosed with type 1 diabetes.
Methods. The Study was conducted in the Pediatric Clinic in Constanta, between January 2011 and May 2015 on a group of 43 children with type 1 diabetes.
We evaluated the presence of the following antibodies at the onset of the disease: antiglutamat decarboxylase (GAD-II), anti-tyrosine phosphatase IA2, anti-insulin antibodies, anti-thyroperoxidase antibodies, anti-thyroglobulin antibodies, anti-transglutaminase antibodies IgA and IgG.
Results. The presence of specific antibodies for diabetes has been evaluated in 20 out of the 43 patients. 2 patients (10%) had 3 positve antibodies, 9 had 2 positve antibodies (45%), 8 patients had 1 positve antibody (40%) and 2 had negative antibodies (10%). Out of the three types of antibodies the most frequent one is antiglutamat decarboxylase (GAD-II) 14 cases. Thyroid screening: 9 patients (20,9%) tested positive for at least 1 antibody at the onset of diabetes and 1 patient eventually developed thyroid dysfunction. Only 1 patient had positive anti-thyroperoxidase antibodies without developing, until now, thyroid dysfunction. Celiac disease was found in 3 of the the subjects (6.96 %). 1 presented initially borderline anti-transglutaminase antibodies with positivity to one year of diabetes diagnosis. 2 patients performed upper gastrointestinal endoscopy and duodenal biopsy, finding histological stage Marsh II.
Conclusions
Detection of autoantibodies, since the onset of diabetes, prevent complications caused by delayed diagnosis of the associated diseases.
Keywords: diabetes, child, celiac disease, thyroid, autoimmunity

GASTROENTEROLOGIE / GASTROENTEROLOGY

6. PREVALENTA SI RATA DE ERADICARE A INFECTIEI CU H PYLORI LA COPIII SIMPTOMATICI – STUDIU UNICENTRIC

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Premise: Majoritatea studiilor recente subliniaza declinul prevalentei si a ratei de eradicare a infectiei cu H pylori.
Obiective: Evaluarea dinamicii prevalentei si a ratei de eradicare a infectiei cu H pylori la copiii simptomatici diagnosticati in perioada 2001-2014.
Metode: Studiu prospectiv si retrospectiv a 1552 copii simptomatici (984 fete, intre 6 luni-18 ani) evaluati esofagogastroduodenoscopic in clinica noastra. Infectia activa cu H pylori a fost documentata prin testul ureazei, examen histopatologic si in cazuri selectate prin antigen fecal, cultura si reactie de polimerizare in lant. Au fost comparate 2 loturi de pacienti: grupul A-294 copii evaluati in primii 3 ani (2001-2003) si grupul B-335 pacienti din ultimii 3 ani (2012-2014).
Copiii infectati au fost tratati cu una din terapiile triple standard sau secventiale, celor cu infectie persistenta recomandanduli-se terapii de "salvare".
Rezultate: Infectia activa cu H pylori a fost deocumentata in 791/1552 cazuri (50,96%), cu o prevalenta anuala intre 34,42%-69,87%, fara diferente semnificative intre grupul A/initial (43,87%) si B/final (45,37%). Gena cag A a fost evidentiata la 96/145 copii infectati. Rata de eradicare a infectiei cu H pylori a fost de 71,93% (569/791) cu o scadere de la 83,91% in 2001, la 70,83% in 2014 si eficienta superioara a terapiei secventiale (84,20%) versus tripla terapie (74,5%).
Concluzii: Rata infectiei cu H pylori la copiii studiati (50,96%) nu confirma declinul recent semnalat in tarile dezvoltate. Cresterea prevalentei H pylori in grupul B fata de grupul A se justifica prin factorii epidemiologici, socioeconomici si caracterul selectiv, impunand studii multicentrice.
cuvinte cheie: helicobacter pylori, copii simptomatici, prevalenta, tratament

THE PREVALENCE AND ERADICATION RATES OF HELICOBACTER PYLORI INFECTION IN SYMPTOMATIC CHILDREN - A SINGLE CENTER STUDY

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Background: Most of the studies has revealed a decline of the prevalence and eradication rates of H pylori infection.
Objectives: To assess the prevalence and eradication rates of H pylori among symptomatic children diagnosed between 2001-2014.
Methods: A retrospective and prospective study of 1552 symptomatic children (984 girls, 6 months-18 years) who underwent esophagogastroduodenoscopy in our clinic over a 14 years. Active H pylori infection was documented by urease test, histopathology and in selected cases by stool antigen , culture and polimerase chain reaction. Two groups of patients were compared: group A-294 children diagnosed during the first 3 years (2001-2003) and group B-335 patients diagnosed in the last 3 years (2012-2014). Infected children received one of the standard three first-line therapies/sequential regimen.
Results: Active H pylori infection was documented in 791/1552 cases (50,96%), with an annual prevalence rate between 34,42% to 69,87%, without significant differences between group A/earlier (43,87%) and group B/latter (45,37%). The cag A gene was positive in 96/145 infected children. H pylori infection was eradicated in 569/791 cases (71,93%) with a decrease from 83,16% (2001) to 70,83% (2014), with the best results for the sequential regimens (84,20%) versus the triple therapies (74,50%).
Conclusions: The prevalence rate of H pylori in our study (50,96%) is not correlated with the recent decline of H pylori, observed in developed countries. The increase of H pylori prevalence observed in the group B can be justified by epidemiological, socioeconomic factors and the selectivity of the study, that imposed a multicenter study.
key words: Helicobacter pylori, symptomatic children, prevalence, treatment

7. REZULTATE ALE PH-METRIEI ASOCIATE CU IMPEDANTA ELECTRICA ESOFAGIANA LA COPIII CU PH-METRIE NEGATIVA

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Obiectiv: Analiza refluxului non-acid la copiii cu suspiciune de reflux gastroesofagian (RGE) cu pH-metrie normala si implicatiile sale terapeutice.
Material și metoda: La pacientii cu suspiciune de RGE, netratati, investigati prin pH-metrie asociata cu impedanta electrica esofagiana 24 ore, cu pH-metrie normala, s-au analizat numarul episoadelor de reflux usor acid (RUAc), numarul episoadelor de reflux usor alcalin (RUAl), corelatia simptomatica intre reflux si simptom prin analiza probabilitatii de asociere simptomatica, extinderea episoadelor de reflux la nivelul esofagului proximal exprimat in procente.

Rezultate: 20 de copii (13 de sex masculin, 7 de sex feminin), cu varsta între o luna si 11 ani au fost identificati. Motivele pentru care s-a recomandat pH-metria asociată cu impedanta electrica esofagiana au fost: varsaturi recurente si falimentul cresterii în 8 cazuri, wheezing recurent asociat cu varsaturi cronice in 7 cazuri si wheezing recurent in 5 cazuri. Toti pacientii au avut extinderea majoritatii episoadelor de reflux la nivelul esofagului proximal, 16 au avut număr crescut de RUAc, 4 corelatie simptomatica pozitiva si 4 atat numar crescut de RUAc cat si corelatie simptomatica pozitiva. Numarul episoadelor RUAl a fost nul la toti copiii. Recomandarile terapeutice au vizat: dieta hipolaergenica in 7 cazuri, tratament simptomatic la 14 pacienti si tratament antisecretor in 6 cazuri.
Concluzii: Asocierea impedantei electrice pH-metriei aduce noi criterii in stabilirea diagnosticului de RGE patologic si permite o abordare terapeutica corecta a acestor copii.
Cuvinte cheie: reflux gastroesofagian non acid, copii

RESULTS OF COMBINED ESOPHAGEAL IMPEDANCE-PH MONITORING IN SYMTOMATIC CHILDREN WITH NORMAL PH METRY

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Objective: Non-acid reflux analysis in children suspected of gastroesophageal reflux (GER) with normal pH-metry and its therapeutic implications
Materials and methods: For pacients suspected of GER, without treatment, examined with pH-metry combined with 24h multichannel intraluminal impedance, normal pH-metry, the following were analysed: the number of weakly acidic reflux (WAR) episodes, the number of weakly alkaline reflux (WAlkR) episodes, the symptomatic correlation between reflux and symptom by analysing the probability of symptomatic association, the extension of reflux episodes within level of proximal oesophagus recorded in percentage.
Results: 20 children (13 male, 7 female), age between 1 month and 11 years, were recorded. The reasons for recommending pH-metry associated with electrical impedance were recurrent vomiting and growth failure in 8 cases, recurrent wheezing associated with chronic vomiting in 7 cases and recurrent wheezing in 8 cases. All patients showed the extension of the majority of reflux episodes at proximal oesophagus level, 16 showed increased WAR, 4 positive symptomatic correlation and 4 increased WAR as well as positive symptomatic correlation. WAlkR episodes were absent in all children. Therapeutic recommendations: hypoallergenic diet for 7 cases, symptomatic treatment for 14 patients and antisecretor treatment for 6 cases.
Conclusion: Association of electrical impedance to pH-metry provides new criteria in establishing pathological GER diagnosis, and allows an accurate therapeutic approach of these children.
Key words: non acid gastroesophageal reflux, children

8. VALOAREA UNUI SCREENING BAZAT PE CHESTIONAR PRIVIND IDENTIFICAREA COPIILOR CU RISC CRESCUT DE BOALĂ CELIACĂ

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Identificarea persoanelor cu risc ridicat de boala celiaca (BC) este o condiție esențială pentru diagnostic, înregistrând o rată crescută a morbidității datorită posibilelor complicații. Scopul prezentului studiu a fost de a identifica copiii cu risc ridicat de BC cu ajutorul unui chestionar. Material și metode: Studiul a început în noiembrie 2012 și a inclus 1839 de copii cu vârste cuprinse între 3 și 18 ani, din Iași, România. Chestionarului s-a atașat consimțământul informat, și a fost aplicat în 5 grădinițe, 2 școli primare și gimnaziale și 2 licee. Chestionarul conține 15 întrebări relevante, privind semnele și simptomele de BC. Eficacitatea acestei metode a fost calculată a fi de peste 85% pentru persoanele cu un scor mai mare de 24 de puncte (Toftedal P, 2010). În conformitate cu răspunsul, fiecare întrebare a primit între 0 și 4 puncte. Un scor total mai mare de 24 de puncte indică o persoană cu risc crescut pentru BC. Rezultate: Doar 35.88% dintre chestionarele distribuite au fost incluse în analiza finală (891 persoane - 48.45%, a returnat chestionarul și 660 de persoane au completat corect chestionarul). 12 copii - 1,81%, au avut peste 24 de puncte. Majoritatea copiilor identificați cu risc crescut de BC au vârste între 15 și 17 ani. Acești copii vor fi investigați în continuare pentru a certifica diagnosticul BC. Concluzii: Prevalența BC poate fi mai mare decât se cunoaște de fapt. Prin urmare, acest chestionar poate fi util ca un instrument de screening în depistarea persoanelor cu risc crescut de BC.
Cuvinte cheie: boală celiacă, risc ridicat, screening, chestionar, copil

THE VALUE OF A QUESTIONNAIRE BASED SCREENING IN IDENTIFICATION OF CHILDREN AT HIGH RISK FOR CELIAC DISEASE

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Identifying the persons with high risk of celiac disease (CD) is a critical value for the diagnosis which has a high morbidity rate due to a lot of complications. The aim of the present study was to identify the children with a high risk of CD using a questionnaire. Material and methods: The study started in November 2012 and included 1839 children between 3 and 18 years-old, from Iasi, Romania. We applied the questionnaires contained the "*Informed Consent*"in 5 kindergartens, 2 primary and middle schools, 2 highschools. The questionnaire contained 15 relevant questions on CD signs and symptoms. The efficacy of this method was calculated to be over 85% for persons with a score higher than 24 points (Toftedal P, 2010). According to the answer, each question received 0 to 4 points. A total score of more than 24 points indicates a person at high risk for CD. Results: Only 35.88 % of the distributed questionnaires were included in the final analysis (891 persons - 48.45 %, returned the questionnaire and 660 persons had correctly completed it). 12 children – 1.81 %, achieved more than 24 points. Most children identified to have a high risk for CD are between 15 and 17 years old. These children will be further investigated in order to certify the CD diagnosis. Conclusions: The prevalence of CD may be higher than we actually know. Therefore, this questionnaire can be useful as a screening test in detecting persons with high risk for CD.

Key words: celiac disease, high risk, screening, questionnaire, child.

9. REFLUXUL GASTROESOFAGIAN LA NOU NASCUT

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Refluxul gastroesofagian (RGE) este una dintre cele mai frecvente patologii in perioada neonatala. RGE se datoreaza imaturitatii sfincterului gastroesofagian inferior, care determina trecerea continutului gastric in esofag.
Scopul lucrarii: identificarea refluxului gastroesofagian la nou nascutul cu regurgitatii si varsaturi.
Material si metoda: 42 de copii au prezentat regurgitatii si varsaturi din cei 110 nou nascuti care s-au prezentat in perioada noiembrie 2014- aprilie 2015 la Spitalul Clinic de Urgenta pentru Copii "Sf. Ioan" Galati, Compartimentul Neonatologie, RGE s-a confirmat la 7 nou nascuti (tranzitul gastroesofagian)
Rezultate: Debutul varsaturilor a fost in primele doua saptamani de viata, 5 cazuri (71, 4%/ 28, 57%) au fost de sex feminin, 3 cazuri (42, 8%) crestere deficitara, 1 caz (14, 28%) malformatie congenitala de cord, 1caz (14, 28%) manifestari respiratorii, 1 caz (14, 28%) stenoza hipertrofica de pilor. Tratamentul a fost nonfarmacologic (pozitie antireflux, micsorarea intervalului orar intre mese 10- 12 pranzuri, evitarea manevrarii 30 de minute). 1 caz (14, 28%) a primit formula de lapte antireflux, in 2 cazuri (28, 57%) s-au asociat inhibitori de pompa de protoni, in 4 cazuri(57, 14%) supresori ai refluxului.
Concluzii: Majoritatea au raspuns la tratamentul nonfarmacologic.
Cuvinte cheie: reflux gastroesofagian, stenoza hipertrofica de pilor, varsaturi

GASTROESOPHAGEAL REFLUX IN NEWBORNS

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Gastroesophageal reflux is one of the most common digestive entities encountered in the neonatal period. It arises due to the immature inferior gastroesophageal sphincter, which allows the gastric contents back into the esophagus.
Objective: To identify the gastroesophageal reflux in newborns that presented vomiting and regurgitations.

Material and method: 110 newborns were presented to the Neonathology Compartment in the Clinical Emergency Hospital for Children "Sfantul Ioan" Galati, 42 of which were experiencing vomiting and regurgitations. Out of the 42 newborns, 7 were diagnosed with gastroesphageal reflux after an upper gastrointestinal series (barium meal) was performed.
Results: vomiting episodes debuted at 2 weeks after birth, 5 were female newborns (71.4%/28.57%), 3 cases (48%) were accompanied by slow growth, 1 by congenital heart malformation (14.28%), 1 by respiratory manifestations (14.28%), 1 by hypertrophic pyloric stenosis (14.28%). The treatment applied was nonpharmacologic (anti-reflux positioning, small frequent 10-12 meals, avoiding manipulation for 30 minutes after the meal), antireflux milk formula, but proton pump inhibitors were associated in 2 cases (28.57%), and anti-reflux medication in 4 cases.

10. BOALA DIAREICA LA SUGAR – DEBUTUL UNEI BOLII RARE

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INTRODUCERE:Boala diareica la sugar prezinta o paleta larga de cauze, bolile rare numarandu-se printre cele mai greu de diagnosticat.PREZENTARE:Gemeni in varsta de 4 luni, se prezinta in Clinica de Pediatrie cu: caune diareice trenante, subfebrilitati.Din istoricul bolii, remarcam 2 internari anterioare in ultima luna in Clinica de Boli Infectioase pentru o simptomatologie similara:scaune diareice cu sange si mucus, febra, infectie urinara cu E. Coli.În istoric, mama relateaza aparitia aftelor bucale in jurul varstei de 2 luni si jumatate. Investigatiile paraclinice efectuate in prima etapa au evidentiat teste fecale ELISA pozitive pentru Adenovirus si Rotavirus.Hemoleucogramele urmarite in dinamica au dezvaluit o limfopenie in continua scadere, cu valori normale ale celorlalte globule albe, insotita de anemie si trombocitoza reactiva, suspicionandu-se o imunodeficienta. Analiza subpopulatiilor limfoctiare a evidentiat un numar scazut de limfocite T si celule Natural Killer Coreland toate datele anamnestice, clinice si paraclinice s-a emis diagnosticul de Imunodeficienta combinata severa(SCID), ce a fost ulterior confirmat genetic.Pacientii au beneficiat de transplant de maduva alogenic, cu evolutie favorabila ulterioara la un caz, celalalt decedand. CONCLUZII:Imunodeficiența combinată severa(SCID) este un sindrom caracterizat prin infecții recurente, diaree, dermatită și retard de creștere.Reprezintă prototipul imunodeficiențelor primare și este cauzată de numeroase defecte moleculare care duc la compromiterea severă a numărului și funcției limfocitelor T, limfocitelor B și a celulelor Natural Killer Din punct de vedere clinic, majoritatea pacientilor prezinta un debut de boala inainte de varsta de 3 luni.Fara o diagnosticare corecta si prompta, SCID conduce la infectii severe si deces pana la varsta de 2 ani.

Cuvinte cheie: sugar, diaree, SCID

GENETICĂ / GENETICS

11. CORELAȚII GENOTIP-FENOTIP ÎN PEDIATRIE. SERIE DE CAZURI

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Scop. Autorii evidentiaza demersul diagnostic la pacienti cu dismorfism facial, retard mental şi sindrom pluri-malformativ. Metode. Autorii prezintă doi copii cu fenotip sugestiv pentru anomalie genetica. Istoric familial (ambele cazuri): fara relatii de consangvinitate. Cazul 1: sugar de 2 luni (cariotip 46,XY) internat pentru evaluare in contextul ambiguitatii genitale. Examenul clinic: microcefalie; țesut subcutanat slab reprezentat (aceeasi greutate ca la nastere), hipotonie generalizată, dismorfism cranio-facial (fante palpebrale cu dispoziție anti-mongoloidă, ptoză palpebrală, epicantus, nări anteversate, micrognație; urechi rotate posterior), sindactilie degete 2/3 picior bilateral, ambiguitate genitală (micropenis, scrot bifid, hypospadias scrotal). Cazul 2: sugar sex masculin in varsta de 5 luni, internat pentru whezing recurent. Examenul clinic: suprapondere, dispoziție mongoloidă fante palpebrale, politelie; brahidactilie, polidactilie post-axială picior, sindactilie degete 5/ 6 picior stâng. Rezultate. Investigatiile serologice au relevat (cazul 1) valori reduse ale colesterolemiei si testosteronului seric, valori normale 17-hidroxi-progesteron. Imagistic: hipoplazie corp calos, duplicatie renala. Ambiguitatea genitala si malformatiile multiple au determinat testarea 7 si 8-dehidro-colesterolului seric, evidentiind valori crescute ale acestora si confirmand sindromul Smith-Lemli-Opitz. La cazul 2, diagnosticul diferential a inclus sindroamele Bardet-Biedl, Laurence-Moon, Cohen, Alstrom, McKusick-Kaufman, Simpson-Golabi-Behmel, Prader-Willi. Testarea genetica (sugar si parinti) a depistat mutația homozigotă "nonsense" a genei BBS12 (c.1063C>T, p.Arg.355), confirmand sindromul Bardet-Biedl. Ambii

părinți sunt purtători heterozigoți pentru aceeași mutație. Concluzii. 1.Cazurile descrise au prezentat dismorfism facial, retard mental si anomalii genitale, justificand investigatii genetice suplimentare; 2.Cazurile analizate releva importanta analizei minutioase a particularitatilor dismorfice pentru fiecare caz in parte.
Cuvinte cheie: dismorfism, boli genetice, anomalie genitala.

PHENOTYPE – GENOTYPE CORRELATIONS IN PEDIATRICS. CASE SERIES

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Aims. The authors emphasize the necessary steps for diagnosis establishing in children characterized by face dysmorphism, mental retardation and congenital anomalies. Methods. The authors present 2 children with phenotype suggestive for genetic anomalies. The family history (both cases): no related parents. First case: 2 month-old male infant (karyotype 46,XY) admitted for futher evaluation because of genital ambiguity. Clinical exam: same weight as compare to birth weight, microcephaly; hypotonia, down-slanted palpebral fissures, epicanthus, anteverted nostrils, micrognathia; posteriorly rotated ears, syndactyly affecting toes 2/ 3 (bilateral), genital ambiguity (micropenis, scrotal hypospadias). Second case: 5 month-old male infant admitted for recurrent wheezing. Clinical exam: overweight, up-slanted palpebral fissures, polithelia, brachydactyly, postaxial polydactyly (bilateral foot), unilateral syndactyly affecting toes 5/6, hypogenitalism. Results. The blood investigations revealed (first case) low cholesterol and testosterone levels, normal value for 17-hydroxyprogesterone. Imagistic evaluation: kidney duplication, corpus callosum hypoplasia. Because of multiple malformations correlated with hypocholesterolemia, the authors tested the sterol profile that revealed increasing of cholesterol precursors (7 and 8-dehydro-cholesterol) and confirmed Smith-Lemli-Opitz syndrome. For second case, the differential diagnosis included following syndromes: Bardet-Biedl, Laurence-Moon, Cohen, Alstrom, McKusick-Kaufman, Prader-Willi. The genetic evaluation revealed homozygous nonsense mutation in BBS12 (c.1063C>T, p.Arg.355) and established Bardet-Biedl syndrome diagnosis (both parents are heterozygous carriers). Conclusions. 1.The phenotype in both children were characterized by craniofacial dysmorphism, mental retardation and genital anomalies, requiring additional genetic tests; 2.These cases emphasize the importance of close analysis regarding dysmorphic peculiarities for every pediatric patient.
Key words: dysmorphism, genetic disorders, genital anomaly

12. .FIBROZA CHISTICĂ COMPLICATĂ CU ALCALOZĂ METABOLICĂ HIPOCLOREMICĂ: PREZENTARE DE CAZ

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Introducere: Alcaloza metabolică hipocloremică, cunoscută şi sub numele de sindrom pseudo-Bartter (SPB) este o complicație a fibrozei chistice (FC). Obiectiv: Prezentarea cazului unui sugar de 8 luni, sex feminin, diagnosticat cu FC la vârsta de 3 luni, complicat cu SPB. Prezentare de caz: Un istoric pozitiv pentru bronhopneumonii repetate, diaree cronică și falimentul creșterii a determinat efectuarea testului sudorii, care a fost pozitiv (NaCl 110mm / l - sistem Nanoduct). Am efectuat, de asemenea, o analiză genetică moleculară a genei CFTR pentru 29 de mutații (kit Elucigene CF 29). Acesta a relevat un status heterozigot pentru mutația F508del-CFTR. S-a instituit terapia nutrițională, fizioterapia, precum și substituția cu enzime pancreatice. În ciuda tratamentului și a complianței familiei la tratament, curba ponderală a rămas nesatisfăcătoare. La cinci luni se prezintă cu alcaloză metabolică hipocloremică severă, hiponatremie, respectiv hipopotasemie. După reechilibrarea hidro-electrolitică și acido-bazică inițială, am instituit terapia orală cu soluție hipertonă de NaCl, rezultatul fiind îmbunătățirea creșterii. Testul sudorii s-a menținut pozitiv și după îmbunătățirea statusului nutrițional (85mm/l). Concluzie: Testul sudorii rămâne patognomonic pentru diagnosticarea FC. SPB întârzie recuperarea nutrițională.
Cuvinte cheie: fibroza chistică, sindrom pseudo-Barter

CYSTIC FIBROSIS WITH HYPOCHLOREMIC METABOLIC ALKALOSIS: CASE REPORT

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Introduction: Hypochloremic metabolic alkalosis is a complication of cystic fibrosis (CF) also known as Pseudo-Bartter syndrome (PBS). Aim: To present the case of an 8 months baby girl diagnosed with CF at 3 months of age, who presented at 5 months with PBS. Case report: A history of repeated pneumonia, diarrhea and failure to thrive lead to performing a sweat test, which was positive (NaCl 110mM/l – Nanoduct system). We also performed a molecular genetic

analysis of the CFTR gene for 29 mutations (kit Elucigene CF 29). It revealed a heterozygous state for F508del-CFTR. Nutritional therapy and pulmonary therapy was started. The infant was put on an enzyme replacement therapy. Despite the treatment and the family's compliance, her failure to thrive continued. At 5 months she presented with PBS. After the acute phase was resolved, we started supplementing with an oral NaCl solution and the result was an improved growth. The sweat test remained positive after her growth improved (85mM/l). Conclusion: The sweat test remains pathognomonic for diagnosing CF. PBS delays the improvement of the nutritional status.

Keywords: Pseudo-Bartter syndrome, cystic fibrosis

HEMATOLOGIE - ONCOLOGIE / HEMATOLOGY - ONCOLOGY

13. INFECTIA CU PARVOVIRUS UMAN B19 LA PACIENTII CU CANCER

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Introducere:
Infectia cu Parvovirus B19 poate induce la gazdele imunocompromise o criza aplastica cronica reprezentand o provocare de diagnostic si de conduita terapeutica.
Material si metoda:
Autorii prezinta un caz de infectie cu Parvovirus uman B19 la o adolescenta in varsta de 16 ani tratata chimioterapic cu 3 cure VIP (Cisplatinum+Etoposidum+Ifosfamidum+Mesnum) pentru tumora ovariana dreapta cu metastaze peritoneale.
Internata in data de 30.12.2014 pentru a patra cura VIP, se constata paloare marcata a tegumentelor, hemograma evidentiaza anemie marcata (Hb 5,8g/dl), leucopenie, neutropenie, limfopenie, trombocitopenie.
Anemia decelata a fost interpretata initial ca fiind secundara chimioterapiei, copilul a primit repetate transfuzii de masa eritrocitara, hemoglobina mentinandu-se la valori de pana la 7,6 g/dl (in absenta evidentierii unei surse de sangerare si hemolizei).
Aspiratul medular si biopsia medulara efectuata releva – Eritroblastopenie secundara infectiei cu Parvovirus uman B19.
Diagnosticul a fost confirmat de titrul crescut de Ac IgM si IgG antiParvovirus H B19 cat si de prezenta AND-ului virusului B19 cu un nivel de 180.000UI/ml in serul pacientei.
Sub tratament cu imunoglobuline i.v., transfuzii de masa eritrocitara si Filgrastinum evolutia a fost favorabila, permitand administrarea curei a IV-a VIP urmata de 5 zile de terapie cu imunoglobuline i.v..
Concluzii:
Infectia cu Parvovirus uman B19 la pacientii cu cancer afecteaza maduva osoasa cu producerea unei citopenii severe – in special anemie, impunand cel putin temporar intreruperea chimioterapiei.

Cuvinte cheie: cancer, imunodeprimat,Parvovirus.

THE INFECTION WITH HUMAN PARVOVIRUS B19 AT CANCER PACIENTS

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Introduction: Human Parvovirus B19 infection can cause at immunocompromised hosts an aplastic anemia that represents o challenge for diagnosis and making the terapeutical decision.
Material and method:
The autors presents a case of Parvovirus H. B19 infection at an adolescent of 16 years old treated with chemoteraphy, having 3 cures of VIP (Cisplatinum+Etoposidum+Ifosfamidum+Mesnum) for the ovarian neoplasia and peritoneal metastasis.
Hospitalized in 30.XII.2014 for the 4th cure of VIP,we observed the extremely paleness of teguments; the blood tests showed anemia (Hb 5,8g/dl), leucopenia, neutropenia, lymphopenia and thrombocytopenia.
The anemia, initially, was interpreted as secondary to chemotherapy, the child received red blood cell transfusion, the hemoglobin maintained at values of up to 7.6 g /dl (in the absence of a source of bleeding and hemolysis).
Medullary bone marrow aspirate and biopsy performed revealed – red cell aplasia that was secondary to the infection with Parvovirus H. B19.
The diagnosis was confirmed by increased titers and IgG and IgM Antibodies antiParvovirus H B19 and the B19 virus DNA present with a level of 180,000 / ml in serum of patient.

Under the i.v. immunoglobulin, transfusions of packed red blood cells and Filgrastinum the evolution was favorable, allowing the 4th cure of VIP , followed by another 5 days of intravenous immunoglobulin therapy.
Conclusions:
Human Parvovirus B19 infection in cancer patients (immunosuppressed) affects the bone marrow to produce a severe cytopenias - especially anemia, setting at least temporarily the discontinuing of chemotherapy.
Key words: cancer, immunocompromised, Parvovirus

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14. SINDROMUL DE ENCEFALOPATIE POSTERIOARA REVERSIBILA - COMPLICATIE SEVERA A TRATAMENTULUI DE INDUCTIE PENTRU LEUCEMIA ACUTA LIMFOBLASTICA LA COPII

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Scopul si obiectivele studiului. Prezentarea si analiza sindromului de encefalopatie posterioara reversibila ("Posterior Reversible Encephalopathy Syndrome", PRES), complicatie severa aparuta in cursul tratamentului de inductie, la doua paciente cu leucemie acuta limfoblastica (LAL).
Material si metode. Pacientele B.T., 7 ani, cu LAL cu precursori B si P.AM., 8 ani, cu LAL cu celule T mature, fara afectare SNC la debut, au prezentat in cursul tratamentului de inductie (Protocol ALL BFM-2000 - Prednison, Vincristin, Daunoblastin, Asparaginaza, Metotrexat intrarahidian) manifestari neurologice severe pentru care s-a recomandat examinarea cerebrala RMN.
Rezultate si discutii. Manifestarile neurologice aparute in afebrilitate in ziua 27 si respectiv 30 de tratament, precedate in ambele cazuri de pusee de hipertensiune arteriala (HTA), au fost de tip criza focala motorie secundar generalizata urmata de stare de coma. RMN cerebral a evidentiat aspecte tipice pentru PRES: leziuni in hipersemnal T2 si FLAIR de edem vasogenic si citotoxic - cortico-subcorticale bilaterale, simetrice, cu localizare preferentiala parieto-occipitala. Ambele paciente au prezentat post-criza HTA greu controlabila cu tripla asociere de medicatie antihipertensiva. In urma tratamentului prompt instituit (anticonvulsivant, depletiv, antihipertensiv) evolutia in ambele cazuri a fost lent favorabila, dupa aproximativ o luna de la debut, cu remisiune clinica completa si ameliorare semnificativa a leziunilor cerebrale la RMN.
Concluzii. Tratamentul de inductie pentru LAL asociaza factori majori de risc pentru PRES (HTA secundara corticoterapiei, efecte citotoxice directe - Metotrexat intrarahidian, Asparaginaza). Diagnosticul si tratamentul prompt al PRES sunt esentiale pentru prevenirea decesului, a sechelelor majore si remisiunea completa.
Cuvinte cheie: PRES, LAL, HTA

POSTERIOR REVERSIBLE ENCEPHALOPATHY SYNDROME - SEVERE COMPLICATION OF INDUCTION CHEMOTHERAPY FOR ACUTE LYMPHOBLASTIC LEUKEMIA IN CHILDREN

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Scope and objectives. Presentation and analysis of posterior reversible encephalopathy syndrome (PRES), severe complication of the induction chemotherapy treatment, in two patients with acute lymphoblastic leukemia (ALL).
Material and methods. Two female patients - B.T., aged 7 years, ALL with B cell precursors, and P.AM., aged 8 years, ALL with mature T cells, without CNS involvement at diagnosis, developed severe neurological manifestations in the course of chemotherapy induction treatment (Protocol ALL BFM-2000 with prednisone, vincristine, daunoblastin, asparaginase, intrathecal methotrexate), and were investigated through cerebral MRI.
Results and discussions. The neurological manifestations appeared without fever, in the day 27th and respectively 30th of induction treatment, preceded in both cases by hypertensive episodes, and were secondary generalised focal motor seizures followed by coma. The cerebral MRI showed typical aspects of PRES: bilateral symmetrical cortico and subcortical lesions, T2 weighed and FLAIR, of vasogenic and cytotoxic edema, with a preferential posterior parieto occipital location. After this episode, both patients had difficult to control hypertension with three antihypertensive drugs. Under the promt treatment (anticonvulsants, depletion, antihypertensive agents) both cases had a slow good evolution in about one month after PRES was diagnosed, with complete clinical remission and significant improvement of the cerebral lesion on MRI.
Conclusions. The chemotherapy induction treatment in children with ALL associates major risk factors for PRES (hypertension secondary to corticotherapy, cytotoxic effects of intrathecal methotrexate, asparaginase). The prompt diagnosis and treatment are essential for the prevention of death, major sequelae, and complete resolution of PRES.
Key words: PRES, ALL, hypertension

HEPATOLOGIE / HEPATOLOGY

15. HEPATITA ACUTA RECURENTA INDUSA DE ALBENDAZOL – PREZENTARE DE CAZ

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Albendazolul este un medicament antihelmintic cu spectru larg, metabolizat in ficat. Acesta, ca toate medicamentele, poate provoaca modificari ale testelor functionale hepatice, evoluand pana la insuficienta hepatica acuta severa. Hepatita toxica indusa de albendazol este rar raportata in patologia pediatrica.

Prezentam cazul unui adolescent in varsta de 15 ani, mediul rural, internat in Clinica II Pediatrie in martie 2015 pentru un tablou biologic de insuficienta hepatica acuta de etiologie necunoscuta, manifestata prin citoliza hepatica, cu TGO (1626U/L), TGP (2686U/L) si alterarea crazelor sangvine (INR=2.41; TQ=26,6s), cu debut la 14 zile de la administrarea primei doze de Albendazol. Mentionam ca adolescentul a mai avut in antecedente (2010) un sindrom citolitic (TGP=1000UI/L), afirmativ la administrare empirica de Albendazol, care s-a remis dupa incetarea medicatiei.

Pe baza istoricului medical, a examenelor clinice si paraclinice , a normalizarii profilului biochimic hepatic dupa intreruperea albendazolului si tratamentul insuficientei hepatice, utilizand Scala CIOMS/ RUCAM (Roussel Uclaf Causality Asseement Method of the Council for International Organisation of Medical Science) propusa pentru a stabili relatiile de cauzalitate între medicamente si afectarea ficatului , (scor=13 - " definit sau foarte probabil " la scor > 8), s-a stabilit diagnosticul de hepatita acuta toxica indusa de albendazol. Mecanismul exact al hepatitei induse de albendazol nu este cunoscut. Cazul atrage atentia asupra faptului ca noi, ca si clinicieni, trebuie sa fim constienti de aceste reactii adverse rare, dar semnificative, aparute dupa administrarea unor medicamente, consumate cu usurinta .

Cuvinte cheie: hepatita toxica, copil, albendazol

RECURRENT ACUTE HEPATITIS INDUCED BY ALBENDAZOL – CASE REPORT

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Albendazol is a broad spectrum anthelmintic drug metabolized in the liver. It, like all drugs, can cause changes in liver function tests, evolving to severe acute liver failure. Albendazol induced toxic hepatitis is rarely reported in pediatric pathology.

We present a 15 year old teenager, rural, interned in Pediatrics Clinic II in March 2015 for a biological picture of acute liver failure of unknown etiology, manifested by hepatic cytolysis with TGO (1626U / L), TGP (2686U / L) and impaired blood values (INR = 2.41; TQ = 26,6s), with onset at 14 days after the first dose of Albendazole. The teenager had a history (2010) cytolytic syndrome (TGP = 1000 / L), so the empirical administration of albendazol, which resolved after stopping the drog.

Based on medical history, clinical and laboratory examinations, the normalization of liver biochemical profile after stopping albendazol and liver failure treatment, using Scala CIOMS / RUCAM (Roussel Uclaf Causality Asseement Method of the Council for International Organization of Medical Science) proposed to establish relationships causal link between drugs and liver damage (score = 13 - "defined or very likely" to score> 8) was diagnosed of acute hepatitis induced toxic albendazol. The exact mechanism of albendazol-induced hepatitis is yet unknown.This case draws attention to the fact that we, as clinicians, need to be aware of these rare but significant side effects occurred after the administration of drugs often consumed empirically.

Keywords: toxic hepatitis, child, albendazol

16. DEFICITUL DE ALFA 1 ANTITRIPSINA – CAPCANA DE DIAGNOSTIC IN HEPATITA COLESTATICA LA SUGAR

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Introducere: Deficitul de alfa-1 antitripsina (A1AT) este o afectiune genetica, autosomal recesiva, caracterizata prin scaderea nivelului A1AT in ser si tesuturi. A1AT este o proteina de faza acuta. Afectarea hepatica este rara la nou-nascut, iar prognosticul bolii este in majoritatea cazurilor bun.

Prezentare de caz: Raportam cazul unei paciente care la varsta de 2 luni si 2 saptamani prezenta febra inalta, diaree, modificari celsiene la nivelul genunchiului stang cu imobilizare totala. Paraclinic: important sindrom inflamator de tip bacterian, transaminaze crescute (ASAT 525U/l, ALAT 1045U/l), sindrom de colestaza, sindrom hepatopriv, serologie pozitiva pentru infectie acuta cu CMV si EBV, nivelul A1AT normal, examen de scaun pozitiv pentru Rotavirus, urocultura pozitiva pentru E.coli. Ecografia abdominala a fost normala, iar radiografia si ecografia de genunchi au fost sugestive pentru

un proces inflamator articular. S-a stabilit diagnosticul de pielonefrita cu E.coli, artrita septica, enterocolita cu Rotavirus, hepatita CMV si EBV. S-a initiat tratament cu vancominina, gentamicina, antitermice, perfuzii de rehidratare si hepatoprotectoare, cu evolutie clinica favorabila, dar cu transaminaze constant crescute. La control se deceleaza nivelul scazut al A1AT, caracteristic deficitului de A1AT, diagnostic sustinut de testele genetice (status homozigot pentru alela Z).
Concluzii: Este vorba despre un pacient cu deficit de A1AT la care in contextul procesului infectios initial nivelul A1AT a fost normal (reactant de fază acuta), fiind o reala capcana de diagnostic avand in vedere ca au existat asociat si alte etiologii ale hepatitei colestatice.

ALPHA 1 ANTITRYPSIN DEFICIENCY - A TRAP IN THE DIAGNOSIS OF CHOLESTATIC HEPATITIS IN INFANT

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Background: Alpha1 antitrypsin (A1AT) deficiency is an autosomal recessive genetic condition, characterized by decreased A1AT level in the blood and tissue. A1AT is an acute phase protein being particularly influenced by inflammatory mediators. About 10% of infants with A1AT deficiency develop liver disease but the prognosis on the most of these cases is good.
Case report: We are reporting the case of girl who presented at the age of 2 months and 2 weeks with high fever, diarrhoea and inflammatory signs on the left knee. Laboratory tests: elevated acute-phase reactants, high level of transaminases (ASAT 525 U/l, ALAT 1045 U/l), elevated alkaline phosphatase and gamma-GT, prolonged prothrombin time, positive serology for acute infection with cytomegalovirus, Epstein-Barr virus and normal level of A1AT. She also had a Rotavirus infection and urine culture positive for E Coli. Based on the clinical examination and laboratory parameters our diagnosis was E.Coli acute pyelonephritis, Rotavirus enteritis, and cytomegalovirus and Epstein - Barr virus infection. Treatment was with antibiotherapy (vancomycin, gentamicin), antipyretics, rehydration and arginine infusion. The clinical evolution was good but with persistent high serum level of transaminases and we detected at follow-up a low level of A1AT, suggestive for A1AT deficiency. The genetic tests confirmed the diagnosis (homozygous status for Z allele).
Conclusion: We are presenting an A1AT deficiency case with normal A1AT serum level at first admission secondary to infection (as A1AT is an acute-phase reactant). It was a real diagnostic trap due to the association of other causes of cholestatic hepatitis in this patient.

IMAGISTICA / MEDICAL IMAGING

17. IMAGISTICA CREIERULUI LA NOU NASCUT SI SUGAR – CE METODA DE INVESTIGARE ALEGEM?

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Accesibilitatea la metode tot mai performante de investigare a creierului nou nascutului si sugarului a dus la diagnosticarea precoce a unor patologii cerebrale. Scopul lucrarii: evaluarea metodelor de diagnostic imagistic al patologiei cerebrale si a necesitatii utilizarii investigatiilor "in trepte". Material si metoda: Studiu retrospectiv pe o perioada de 2 ani (2013-2015). Acesta a cuprins 1450 de pacienti cu varsta cuprinsa intre 0 si 18 luni, media 3.4 luni. Toti pacientii au fost investigati prin ultrasonografie. Dintre acestia, 200 de pacienti (13.8%) au beneficiat de examinare CT si si un numar mai redus 75 pacienti (5.17%) de investigatie RMN. 40 (2.75%) dintre pacienti au beneficiat de toate cele 3 metode. Rezultate: Majoritatatea pacientilor au fost cu patologie minora; acestia au fost monitorizati ecografic. Pacientii diagnosticati cu hidrocefalie evolutiva au fost investigati prin CT in vederea interventiei chirurgicale, iar 15% au beneficiat si de RMN. 3 pacienti cu suspiciune clinica de AVC au necesitat explorare CT, 2 pacienti cu malformatie vasculara au necesitat toate cele 3 metode, iar un pacient cu tumora cerebrala a efectuat de asemenea toate cele 3 investigatii. Discutii: In ceea ce priveste pacientii diagnosticati cu hidrocefalie, investigatiile suplimentare nu au fost justificate. Concluzii: Majoritatea cazurilor nu au necesitat investigatii suplimentare; consideram ca in cazul hidrocefaliilor este suficienta metoda ecografica atat in diagnosticare cat si in urmarire. Investigatiile suplimentare au un rol important in traumatisme, leziuni ale substantei albe, patologie tumorala si vasculara.
Cuvinte cheie: imagistica, creier, nou nascut

BRAIN IMAGING IN NEONATES AND INFANTS, WHICH METHOD TO CHOOSE?

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The availability of increasingly performant methods for investigating the neonate and infant brain led to early diagnosis of cerebral pathologies. Aims: evaluating the diagnostic imaging of cerebral pathology investigations. Material and methods: A retrospective study was conducted over a period of two years (2013-2015). 1450 patients were included aged 0 to 18 months, average 3.4 months. All patients were investigated by ultrasonography. Of these, 200 patients (13.8%) performed a CT examination and a smaller number of 75 patients (5.17%) an NMR investigation. 40 (2.75%) of patients were evaluated using all 3 methods. Results: Most patients had minor pathology; they were monitored by ultrasound. Patients diagnosed with hydrocephaly were investigated by CT prior to surgery and 15% underwent MRI. 3 patients with clinical suspicion of stroke required CT, 2 patients with vascular malformation were subject to all 3 methods, and one patient with a brain tumor was also examined using all 3 methods. Discussions: Additional investigations were not justified for patients diagnosed with hydrocephaly. Conclusions: Most cases did not require further investigation; we consider that ultrasound examination is sufficient in case of hydrocephaly. Additional investigations have an important role in trauma, white matter lesions, tumor and vascular pathology.

Key words: imagistic, brain, newborn

18. ULTRASONOGRAFIA IN HEPATOPATIA ASOCIATA FIBROZEI CHISTICE

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Introducere: Fibroza chistică(FC) se caracterizează printr-un polimorfism clinic impresionant, fie că se manifestă prin pneumopatia cronică obstructivă si insuficienţă pancreatică, fie că se asociază hepatopatie cronică sau diabet zaharat secundar. Diagnosticarea precoce si monitorizarea corectă a tuturor afecţiunilor are ca scop prelungirea duratei si calităţii vieţii acestor copii. Ultrasonografia este o metoda neivazivă extrem de utilă pentru diagnosticul acestor complicaţii.
Scopul lucrării este de a evalua hepatopatia asociată fibrozei chistice cu ajutorul ultrasonografiei.
Material şi metodă: 158 de pacienţi, aflaţi în evidenţa Centrului National de Mucoviscidoza(Fibroza Chistica) Timisoara au fost evaluaţi ecografic în completarea examenului clinic şi a evaluării biologice, Pentru evaluarea ultrasonografica s-a utilizat scorul Williams iar in unele cazuri a fost utilizata si elastografia hepatica.
Rezultate: Prevalenţa hepatopatiei a fost de 32,27% (51 pacienti). Majoritatea pacienţilor 62,74% (32 pacienţi) au prezentat hepatopatie moderată, un procent de 9,8% au asociat ciroză hepatică multilobulară Concluzii: Frecvenţa hepatopatiei asociate fibrozei chistice este semnificativă. Ultrasonografia este o metodă extrem de eficientă pentru diagnosticul si monitorizarea afecţiunilor hepato-biliare din fibroza chistică. Depistarea precoce a acestei afecţiuni permite instituirea unei atitudini terapeutice adecvate , cu îmbunătăţirea speranţei de viaţă a acestor pacienţi.

ULTRASOUND ASPECT IN CYSTIC FIBROSIS LIVER DISEASE

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Introduction: Cystic fibrosis (CF) is a genetic disease characterized by clinical polymorphism, manifested by pancreatic insufficiency, chronic obstructive pulmonary disease , hepatobilliary impairment or secondary diabetes . Hepatobilliary disorders have an important place in the disease`s outcome and ultrasound is a reliable method used to diagnose these complications CF associated. The purpose of this paper is to present data on the diagnosis and monitoring of diseases of liver and biliary disease. Methods: 174 children were included in the transversal study, and followed at the National Center for Mucoviscidoza (Cystic Fibrosis) Timisoara. Ultarsound examinations were performed, besides clinical assesemnet and biochemical evaluation. Gallbladder diseases (GB) were monitored as follows: microgallbladder (GB atrophy), cholelithiasis , cholecystitis . Results: Disorders of GB were diagnosed in 29.3 % of patients; the most common being GB atrophy (20.6 %). Gallstones were seen in 16 patients (31.3 %, compared to patients with other GB diseases), being diagnosed at early age. Conclusions: The prevalence of biliary disease is significantly associated with cystic fibrosis in children. Ultrasound is a very useful method for the diagnosis and monitoring of hepatobilliary diseases CF associated.

IMUNOLOGIE / IMMUNOLOGY

19. DIFICULTĂȚI TERAPEUTICE LA COPII CU IMUNODEFICIENȚE PRIMARE. CAZURI CLINICE

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Scop. Autorii mentioneaza particularitatile terapeutice la doi copii cu infectii recurente secundare imunodeficientei primare (IDP). Metode. Autorii prezinta 2 cazuri cu frecvente spitalizari datorate infectiilor severe: pacienta de 3 ani cu neutropenie severa si sugar de 10 luni cu sindrom hiper-IgM. Istoric familial: parinti consanguini (primul caz). Istoric: pneumonie recurenta, abces vulvar, otita medie supurata (cazul 1); pneumonie lobara, otita medie supurata recurenta (cazul 2). Examen clinic: cazul 1- deficit ponderal, raluri pulmonare, otita supurata bilaterala. Cazul 2- malnutritie, hepato-splenomegalie, otita purulenta bilaterala, pneumonie. Rezultate. Investigatii primul caz: neutropenie severa, *Proteus spp.*in secretia otica, valori reduse elastaza fecala, test iontoforeza normal. Examenul maduvei osoase si imunofenotiparea au relevat apoptoza precursorilor seriei mieloide. Examenul CT toracic a identificat bronsiectazie. Investigatii cazul 2: fenotip imunologic hiper-IgM (valori scazute IgA/ IgG, valori crescute IgM), fara anomalii maduva osoasa. Examenul secretiei otice a confirmat *Staphylococcus aureus*, pneumococ penicilino-rezistent si *Candida albicans*. Testele genetice in primul caz au exclus sindroamele Shwachman-Diamond si Pearson, anemia Fanconi. Cazul 2: secventierea ADN a eliminat deficitele CD40/ CD40 ligand/ AICDA/ UNG. Terapia a constat, in ambele cazuri, in antibioterapie de lunga durata si spectru larg, terapie antifungica (bazat pe antifungigrama), factor stimulare colonii granulocitare (cazul 1), substitutie cu imunoglobulina umana si profilaxia pneumocistozei (cazul 2). In ciuda terapiei, se remarca recurenta infectiilor severe. Concluzii. 1.Alegerea terapiei adecvate la pacientii cu IDP ramane o provocare pentru clinician; 2.Initierea terapiei empirice antifungice reprezinta o decizie dificila, intrucat criteriile de initiere a terapiei nu sunt clar definite.

Cuvinte cheie: infectie, imunodeficienta, copil,

TERAPEUTIC DIFFICULTIES IN CHILDREN WITH PRIMARY IMMUNODEFICIENCIES. CLINICAL CASES

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Aims. Authors emphasize treatment peculiarities in two pediatric patients with recurrent infections secondary to primary immunodeficiency (PID) with undefined genetic background. Methods. Authors present two cases frequently admitted due to severe infections: 3 year-old girl with severe congenital neutropenia and 10 month-old male infant with hyper-IgM syndrome. Family history: consanguineous parents (first case). Cases history: recurrent pneumonia, vulvar abscess, purulent otitis media (first case); lobar pneumonia, recurrent purulent otitis media (second case). Clinical exam: first case- impaired nutritional status, lung crackles, suppurative otitis media. Second case- malnutrition, hepatosplenomegaly, bilateral purulent otitis, pneumonia. Results. First case investigations: severe neutropenia, *Proteus spp.* in ear pus, reduced fecal elastase level, normal sweat test. Bone marrow exam/ flow cytometry noticed myeloid cells apoptosis. CT scan reveled bronchiectasis. Second case investigations: hyper-IgM phenotype (IgA/IgG low levels, IgM high value) without bone marrow anomalies. Ear pus analysis revealed *Staphylococcus aureus*, penicillin-resistant pneumococcus, *Candida albicans*. First case genetic analysis excluded Shwachman-Diamond/ Pearson syndromes, Fanconi anemia. Second case: DNA sequencing ruled out CD40 deficiency, CD40 ligand deficiency, AICDA/ UNG deficiencies. The both cases therapies consisted in long duration broad-spectrum antibiotic regimens (using antimicrobial susceptibility tests), antifungal agents (using antifungigram), granulocyte colony stimulating factor (first case), immunoglobulin substitution therapy/ pneumocystosis prophylaxis (second case). In spite of therapy, authors noticed severe and recurrent infections. Conclusions. 1.It’s difficult to implement a proper therapeutic management for PID patients; 2.Empirical antifungal therapy continues to be a problematic decision since criteria to start therapy remain poorly defined.

Key words: infection, immunodeficiency, child.

BOLI INFECTIOASE ȘI VACCINOLOGIE/ INFECTIOUS DISEASE AND VACCINOLOGY

20. CRESTEREA REZISTENTEI LA ANTIBIOTICE A PNEUMOCOCULUI SI IMPLICATIILE TERAPEUTICE

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Background. La copii pneumococul (*S.pneumoniae*) poate fi izolat din nazo-faringe (purtatori) si poate fi izolat, ca agent etiologic, in conjunctivite, infectii respiratorii, meningite etc. Alegerea terapiei optime este dificila datorita timpului prelungit necesar obtinerii antibiogramei (ABG), dificultatilor de procesare a acesteia si rezistentei crescute a pneumococului. In Romania vaccinarea anti-pneumococica nu se efectueaza de rutina. Scopuri. 1.Analiza spectrului de rezistenta a *S.pneumoniae;* 2.Stabilirea criteriilor de initiere a terapiei empirice raportat la profilul de rezistenta al pneumococului. Metode. In perioada octombrie 2011-decembrie 2014, s-au efectuat ABG pentru *S.pneumoniae* utilizand carduri Vitek AST-P576. Originea probelor: secretii nazale, secretii conjunctivale, secretii otice, hemoculturi, lichid cerebro-spinal, proba urina. Raportat la concentratia minima inhibitorie (CMI) la penicilina, tulpinile au fost impartite in: sensibile (CMI ≤0.06 mg/l), intermediare (0.06 < CMI< 2 mg/l), rezistente (PRP) (CMI ≥ 2 mg/l). Autorii au folosit pentru ABG: Cefotaxim(CTX), Ceftriaxona(CRO), Eritromicina(E), Levofloxacina(L), Vancomicina(VA), Trimethoprim-Sulfamethoxazole(SXT). Rezultate. Dintre 85 ABG validate, autorii au remarcat rezistenta la penicilina in 50,57% din cazuri, 69,33% rezistenta la E, 69,6% rezistenta la SXT, 32,84% rezistenta la CRO, 22,91% rezistenta la CTX. Toate tulpinile au fost sensibile la L, VA. Concluzii. 1.Autorii remarca rezistenta crescuta la Penicilina, E si STX in arealul geografic; 2. Optiunile terapeutice sunt limitate in infectiile pneumococice; 3.Datorita pattern-ului de rezistenta, autorii restrictioneaza utilizarea macrolidelor si STX in otita medie; 4.In infectiile severe, recomandarile terapeutice raman CTX, CRO, VA; 5.Implementarea masurilor educationale este importanta pentru evitarea antibioterapiei excesive.
Cuvinte cheie: pneumococ, rezistenta microbiana, tratament.

PNEUMOCOCCUS RESISTANCE TO ANTIBIOTICS. DIFFICULTIES REGARDING THERAPEUTIC OPTIONS

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During childhood pneumococccus (*S.pneumoniae*) is identified not only in nasopharynx (carriers), but also as etiological agent in conjunctivitis, respiratory airways infections, meningitis etc. Choosing the optimal therapy is difficult due to prolonged time necessary to antibiotic sensitivity test (AST) achievement, difficulty to perform it, pneumococcus strains growing resistance. In Romania vaccination against *S.pneumoniae* isn't offered by national immunization program. Aims. 1.To analyse resistance spectrum for *S.pneumoniae;* 2.To establish empirical therapeutic decisions according to pneumococcus resistance profile in our county. Methods. During October 2011-December 2014, there were performed AST for *S.pneumoniae* using Vitek AST-P576 and AST-ST01 cards. Samples origin: nasal secretions, conjunctival secretions, ear samples, hemocultures, cerebrospinal fluid, urine culture. Correlated with MIC to penicillin, the strains were divided into sensitive (MIC ≤0.06 mg/l), intermediate (0.06 < MIC< 2 mg/l), resistant (PRP) (MIC ≥ 2 mg/l). Authors took into consideration for AST: Cefotaxime(CTX), Ceftriaxone(CRO), Eritromicine(E), Levofloxacin(L), Vancomycin(VA), Trimethoprim-Sulfamethoxazole(SXT). Results. Among 85 validated AST, authors noticed 50.57% Penicillin resistance, 69.33% E resistant strains, 69.6% SXT resistance, for CRO resistance reached 32.84%, 22.91% CTX resistance. All strains were sensitive for L, VA. Conclusions. 1.Authors observed increased resistance to penicillin, E and STX in our county; 2.Therapeutic options are limited in *S.pneumoniae* infection; 3.According to resistance pattern, authors restrict use of macrolides and STX in otitis media (most frequent etiological agent is *S.pneumoniae*); 4.In severe infections, therapeutic choices are CTX, CRO, VA; 5.Implementation of educational measures is important in order to avoid antibiotic overuse.
Key words: pneumococcus, antimicrobial resistance, therapy.

21. STUDIU COMPARATIV AL S.PNEUMONIAE PROVENIT DIN OTITELE MEDII VS PNEUMONII LA COPIII CU VARSTA DE SUB 5 ANI, IN BRASOV

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Introducere: Romania este una dintre tarile care nu au implementat pe programul National de Imunizare vaccinarea pneumococica. Izolatele de pneumococ din tara noastra au rezistenta inalta la antibiotice aspect raportat in repetate randuri.
Scopul studiului: compararea distributiei serotipurilor de pneumococ precum si rezistenta la antibiotice in cadrul a doua boli mucozale otitele medii si pneumonia comunitara, la copiii sub 5 ani in Brasov.

Pacienti si metoda: studiu de tip prospectiv pe o perioada de 5 ani din 2009-2014 in care au fost inrolati copiii sub 5 ani care au avut diagnosticul de otita medie si cel de pneumonie comunitara.
Rezultate: un numar de 117 copii au fost inrolati dintre care 29 au avut otite medii si 88 cu pneumonie comunitara la acestia efectuandu-se si serotipare. Varsta medie a fost de 25.1+/-24.01 pentru otite si pentru pneumonii de 27.01+/-28.46 (p=0.78). Saturatia in oxigen a fost mai mica ($p<0.05$) la cei din grupul cu pneumonie. Serotipul 19F a fost mai frecvent izolat din grupul cu otite medii, 23F a fost izolat mai frecvent din grupul cu pneumonii, 6B si 6A mai frecvent in grupul cu pneumonii. Rezistenta la antibiotice a fost inalta la ceftriaxon MIC pentru otite a fost de 48µg/ml iar pentru pneumonii a fost de 64 µg/ml.
Concluzii: in ambele afectiuni mucozale copiii frecvent afectati sunt cei care locuiesc in familii numeroase. Cele mai frecvente serotipuri sunt 14,6B, 19F si 23F. Rezistenta la antibiotice este inalta. Acoperirea cu PCV13 este de 89.6% pentru otite si de 81.8% pentru pneumonii.

COMPARISON STUDY OF S.PNEUMONIAE IN OTITIS MEDIA VS. PNEUMONIA AT CHILDREN UNDER THE AGE OF 5, IN BRASOV COUNTY

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Background: Romania is one of the few countries that have not yet implemented pneumococcal vaccine into the National Immunization Program. Pneumococcal isolates from our country have a high resistance pattern towards antibiotics, that was repeteadly reported.
Aim of the study: was to compare pneumococcus serotype distribution as well as the antibiotic resistenace pattern of it in two mucosal diseases acute otitis media and community acquiered pneumonia at children under the age of 5 in Brasov.
Pacients and method: prospectiv study over a five years period between 2009-2014 in which we enrolled children under 5 years of age with the diagnose of acute otitis media and community acquiered pneumonia.
Results: 117 children were enrolled 29 had otitis media and 88 community aquiered pneumonia, all of these pneumococcal strains being serotyped. Mean age at admittance was for the otitis media group 25.1+/-24.01, while for pneumonia 27.01+/-28.46 (p=0.78). Mean oxygen saturation was lower in pneumonia group ($p<0.05$). 19F was more frequently encountered in the otitis media group while23 F,6A and 6B in pneumonia group. MIC for ceftriaxon in otitis media group was 48µg/ml and in pneumonia group 64 µg/ml.
Conclusions: in both mucosal diseases children that live in crowded homes are more affected. Most frequently encountered serotypes are 14, 6B, 19F and 23F. We have a high antibiotic resistance pattern. Vaccine coverage for PCV 13 is 89.6% for otitis media and 81.8% for pneumonia.

NEFROLOGIE / NEFROLOGY

22. DIAGNOSTICUL PRECOCE SI IMPORTANTA SCREENENG-ULUI GENETIC IN MALFORMATIILE RENO-URINARE LA COPII SI ASCENDENTI

Lazar Viorica, Lazar Dorin, Dumitra Simona, Golumbeanu Mihaela, Horga Adeline Larisa, Covaci Elena Claudia
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Patologia reno-urinara la copil ocupa locul al treilea dupa cea respiratorie si digestiva. Diagnosticul precoce al anomaliilor reno-urinare la copil, asociate cu infectie urinara, permite tratamentul tintit si evitarea cicatricilor renale, precum si abordarea chirurgicala precoce a malformatiilor reno –urinare si prevenirea instalarii insuficientei renale. Aparitia fenotipurilor diferite si a agregarii familiale, demonstreaza ca exista cauze genetice pentru malformatiile reno-urinare. Caracterul complex al malformatiilor reno-urinare este subliniat de variabilitatea fenotipica printre membrii familiei, cu acelasi defect pe o singura gena, cu exprimare variabila, de la anormalitati structurale simptomatice pana la insuficienta renala cronica. Exista posibilitatea ca variatia genetica sa afecteze inflamatia renourinara si sa genereze fibroza interstitiala dupa un episod de pielonefrita acuta. Exista dovezi clare de asociere intre formarea de cicatrici renale la copiii cu reflux vezico-ureteral si polimorfism de inalta conversie a genelor implicate in malformatii renourinare. O cunoastere a patogenezei pentru CAKUT va imbunatati strategiile pentru diagnosticul precoce, follow-up, tratament. Dezvoltarea cercetarilor in

domeniul molecular, implicate in dezvoltarea rinichilor va contribui la dezvoltarea de noi metode de diagnostic si in acelasi timp va permite noi abordari terapeutice.

23. ENZIMELE PLASMATICE ÎN URINĂ LA COPII CU GLOMERULONEFRITĂ ACUTĂ SINDROM NEFROTIC

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Scopul studiului: Aprecierea enzimelor plasmatice în urină şi rolul clinico-evolutiv la copii cu glomerulonefrită acută sindrom nefrotic.

Material şi metode: Studiul se axează pe evaluarea activităţii enzimelor plasmatice în urină la etapele clinico-evolutive la 40 copii cu glomerulonefrită acută (GNA) sindrom nefroticîn stadiul funcţional-compensator. Grupul de control a constituit 20 de copii practic sănătoşi.

Rezultate:

Analiza rezultatelor a determinat o majorare semnificativă a activității y-GTP în urină la pacienții cu sindrom nefrotic de 3 ori în raport cu grupul-martor (0,08 ± 0,003 nmol/s.g. creatinină).

Remarcăm că la pacienţii cu GNA sindrom nefrotic evoluţie trenantă activitatea fosfatazei alcaline în urină a sporit de 2,8 ori (0,36 ± 0,025 nmol/s.g. creatinină, p<0,01), comparativ grupului de pacienţii cu sindrom nefrotic unde activitatea fosfatazei alcaline a sporit de 1,7 ori (0,22 ± 0,003 nmol/s.g. creatinină, p<0,01) în raport cu valorile normale (0,13 ± 0,020 nmol/s.g. creatinină).

În perioada remisiei clinice activitatea fosfatazei alcaline în urină rămâne a fi majorată la pacienții cu GNA sindrom nefrotic, evoluţie trenantă, comparativ grupului martor.

Concluzii:

Monitorizarea activităţii enzimatice în urină reflectă intensitatea procesului inflamator, gradul de alterare a membranelor celulare, topografia leziunilor, permit prognozarea complicaţiilor şi specificarea diagnostică a diferitor variante de glomerulonefrită primară la copil.

URINARY PLASMATIC ENZYMES IN CHILDREN WITH ACUTE GLOMERULONEPHRITIS NEPHROTIC SYNDROME

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Aim: To assess the plasmatic enzymes in urine and to appreciate their clinical role in children with acute glomerulonephritis nephrotic syndrome.

Methods and material: This study evaluates the plasmatic enzymes activity in urine at different clinical-evolutional stages in 40 children with acute glomerulonephritis (GNA) nephrotic syndrome in functional-compensatory stage. The control group includes basically healthy 20 children.

Results: data analysis of the study determined a significant increase of γ-GTP activity in urine, in patients with nephrotic syndrome, 3 times compared to the control group (0,08±0,003 nmol/s.g. creatinine).

We remark that in patients with GNA nephrotic syndrome with trenant evolution, the alkaline phosphatase activity in urine increased 2,8 times (0,36±0,025 nmol/s.g. creatinine, p<0,01)compared to patientswith nephrotic syndrome group which alkaline phosphatase activity increased 1,7 times (0,22±0,003 nmol/s.g. creatinine, p<0,01) compared to normal values (0,13±0,020 nmol/s.g. creatinine).

During clinical remission, the alkaline phosphatase activity in urine remains to be increased in patients with GNA nephrotic syndrome trenant evolution, compared to the control group.

Conclusions: The monitoring of enzyme activity in urine reflects the intensity of the inflammatory process, the degree of alteration of cellular membranes, the topography of lesions, also it allows to predict the complications and to specify the diagnostic peculiarities of different variants of primary glomerulonephritis in children.

NEONATOLOGIE / NEONATOLOGY

24. IMPORTANTA SPECTROSCOPIEI IN INFRAROSU PENTRU MONITORIZAREA OXIGENARII CEREBRALE IN TERAPIA INTENSIVA NEONATALA

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Introducere:
Nou-nascutii prematuri sunt pacienti cu risc din Terapia Intensiva Neonatala (TINN). In ultimii ani, numarul prematurilor extrem de mici, recuperati, a crescut foarte mult. Abordam aceasta problema din perspectiva riscului crescut de insulta hipoxic ischemica perinatala.
Material si metoda:
Studiul s –a desfasurat in Clinica de Neonatologie Bega Timisoara in perioada 01.01.2012 – 01.07.2014. Am inclus 359 de prematuri cu VG ≤32 de saptamani, Gn= 400- 2100 grame. Inregistrarea oxgenarii cerebrale (rSO2) s –a efectuat din primele 4 ore de viata, pana la 72 de ore. In paralel am evaluat parametrii de monitorizare clinico-biologica utilizati de rutina. Pentru monitorizarea rSO2 am utilizat un monitor ce are ca principiu de functionare spectroscopia in infrarosu (NIRS).
Rezultate si discutii:
Monitorul NIRS reflecta modificarea balantei dintre cerere si oferta de oxigen. Pragul critic: scadearea rSO2 >20% din nivelul bazal stabilit, rSO2 fiind indicator al perturbarii oxigenarii cerebrale. Prin AnalyticsTool am interpretat inregistrarile, am calculat fractia de extractie tisulara, valoarea medie si zonele de sub curba cu potential maxim de risc. Monitorizarea rSO2 in paralel cu oxigenarea periferica prin pulsoximetrie, poate ghida administrarea de oxigen. Limite stabilite pentru rSO2 au fost 50 - 80%.
Concluzii:
In TINN este nevoie de metode neinvazive de investigare. NIRS este o metoda moderna de urmarire in timp real a schimbarilor perfuziei tisulare. Valorile inregistrate trebuie interpretate in contextul clinic. NIRS poate ghida administrarea de oxigen in timpul resuscitarii neonatale. Valorile rSO2 ne-au orientat in conduita terapeutica.
Cuvinte cheie: prematuritate, oxigenare cerebrala, spectroscopie in infrarosu, hipoxie.

NEAR INFRARED SPECTROSCOPY FOR CEREBRAL OXIGEN MONITORING IN THE NEONATAL INTENSIVE CARE UNIT

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Introduction:
Premature newborns are a risk category of patients in the Neonatal Intensive Care Unit (NICU). Lately, the number of recovered extremely premature infants has increased. We approach this issue from the hypoxic ischemic injury increased risk point of view.
Material and method:
The study was conducted between 1st of January 2012 – 1st of July 2014. We included 359 premature infants, GA ≤32 weeks, BW= 400- 2100 grams. We monitored cerebral oxygenation (rSO2) from the first 4 hours of life, until 72 hours. We monitored vital functions and blood tests that we routinely perform in the NICU. For rSO2 monitoring we used a device that functions using near infrared spectroscopy (NIRS).
Results and discussions:
NIRS device reflects the balance between the demand and the supply of oxigen. The decrease of rSO2 over 20% from the baseline value is considered critical. rSO2 indicates the disturbance of cerebral oxigenation. Using Analytics Tool we interpreted the recorded graphics, we calculated the tisular extracted fraction of oxigen, the average value and the area under the curve (maximum risk). We used pulsoximetry for peripheral oxigenation. The lower and upper limits for rSO2 were set at: 50 - 80%.
Conclusions:
In the NICU we need a noninvasive method of monitoring cerebral oxygenation; NIRS is a modern and real time method. The records have to be interpreted in the clinical context. NIRS can guide us during ressuscitaion. rSO2 values guided us during critical care of the monitored patients.
Key words: prematurity, cerebral oxygenation, near infrared spectroscopy, hypoxia

25. MONITORIZAREA INVOS A AFECŢIUNILOR GASTRO-INTESTINALE SEVERE LA NOU-NĂSCUT

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Introducere: Ipoteza studiului presupune că alterările oxigenării splanhnice este calea finală comună în patogeneza enterocolitei ulcero-necrotice (EUN). Monitorizarea INVOS detectează modificările fluxului sangvin regional şi ischemia mezenterică locală.
Obectiv: Autorii au evaluat patologic severă ischemică gastro-intestinală la nou-născut utilizând parametrii biologici INVOS şi criteriile clinice folosite pentru monitorizare. Scopul studiului a fost detectarea precoce a modificărilor ischemice la nou-născutul cu anomalii digestive.
Material. Metodă: Un număr de 30 de nou-născuţi cu semne clinice de patologie digestivă severă u intoleranţă digestivă, cu vârstă de gestaţie (VG) de 25-38 săptămâni, greutate la naştere (GN) 750-3250g au fost monitorizaţi pe o durată de 18 luni. Nou-născuţii au fost diagnosticaţi cu malformaţii congenitale cardiace (MCC), EUN, RCIU şi sepsis.
Rezultate: Indexul de oxigenare tisulară a patului vascular este modificat atunci când fluxul sangvin diminuă. Raportul CSOR (oxigenare cerebrală/splanhnică) arată ischemia. Media valorilor CSOR obţinute pentru 6 nou-născuţi cu MCC a fost de 0,42-0,53, în 12 cazuri de EUN 0,57-0,62, 68-69, în RCIU 0,76-0,83 iar în 6 cazuri de septicemie 0,67-0,93. CSOR s-a corelat statistic semnificativ cu GN (p=0,02), VG (p=0,01), tensiunea arterială medie (p=0,01), pH (p=0,01), cantitatea de lactaţi (p=0,01) şi nivelul hemoglobinei (p=0,012).
Concluzie: Monitorizarea INVOS poate semnala stadii precoce ale scăderii perfuziei intestinale, faze iniţiale similare EUN.

INVOS MONITORING OF SEVERE GASTROINTESTINAL DISORDERS OF THE NEWBORN

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Introduction: The hypothesis of the study assumes that alterations of the splanchnic oxygenation is the final common pathway in necrotizing enterocolitis (NEC) pathogenesis. INVOS monitor detects changes in the regional blood flow and local mesenteric ischemia.
Objective: The authors evaluated severe gastrointestinal ischemic pathology in newborns using INVOS, biological parameters and clinical criteria used for monitoring. The aim of the study was early detection of ischemic changes in newborns with digestive disturbances.
Material. Method: 30 newborns having clinical signs of severe digestive pathology with gastric intolerance, gestational age (GA) 25-38 weeks, birth weight (BW) of 750–3250gr were monitored during 18 months. The newborns were diagnosed with congenital heart defects (CHD), NEC, IUGR, and sepsis.
Results: Tissue oxygenation index of the vascular bed is modified when blood flow is reduced. The report CSOR (cerebral/splanchnic oxygenation) shows splanchnic ischemia. The average CSOR values obtained for 6 neonates with CHD cardiac malformations were between 0,42-0.53; in 12 cases of NEC - 0,57-0.62, 68-69; in IUGR - 0,76-0.83; and in 6 cases of septicemia - 0,67-0.93. CSOR had a statistically signifiucant correlation with BW (p=0.02), GA (p=0.01), mean arterial pressure (p=0.01), pH (p=0.01), lactate (p=0.018), hemoglobin levels <10g/dl (p=0.012).
Conclusions: INVOS monitoring may releave early stages of decreased intestinal perfusion, initial phases similar to NEC.

NEUROLOGIE SI PSIHIATRIE INFANTILA / CHILD NEUROLOGY AND PSYCHIATRY

26. PSEUDOTUMOR CEREBRI (HIPERTENSIUNEA INTRACRANIANA IDIOPATICA): CAUZA DE CEFALEE SI TULBURARE A VEDERII LA COPIL

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Introducere: Hipertensiunea intracraniana idiopatica (benigna), numita si pseudotumor cerebri, este o afectiune neurologica caracterizata prin cresterea presiunii intracerebrale in absenta unei formatiuni inlocuitoare de spatiu sau a unei alte afectiuni cerebrale. Este o afectiune care determina pierderea treptata a vederii la copii si adolescenti. Diagnosticul este in cea mai mare parte sugerat de tabloul clinic, dar probele de laborator si investigatiile imagistice sunt necesare pentru confirmarea diagnosticului.
Prezentare de caz: Prezentam cazul unei fetite in varsta de 6 ani diagnosticata cu Pseudotumor cerebri. Simptomatologia a debutat cu 5 zile anterior internarii cu cefalee, diplopie si strabism convergent bilateral. Tomografia computerizata si RMN cerebral nu au decelat modificari patologice. La examinarea oftalmologica presiunea intraoculara si reflexele papilare au fost normale, insa la examinarea fundului de ochi s-a decelat proeminenta papilei optice la ambii ochi, cu contur fluu delimitat, hemoragii peripapilare si vene tortuoase realizand aspectul de "floare de crizantema". De asemenea pacienta prezenta asociat paralizie de nerv cranian VI la nivelul ochiului stang cu limitarea miscarii de abductie a globului ocular. A primit tratament cu depletive cerebrale (manitol) si corticoterapie (dexametazona, prednison) cu evolutie favorabila. O luna mai tarziu aspectul fundului de ochi era normal, cu rezolutia totala a edemului papilar si disparitia simptomatologiei.
Concluzie: Este un caz de pseudotumor cerebri asociat cu paralizia de nerv VI, la care nu s-a gasit nicio cauza infectioasa, toxica, endocrina, metabolica sau sistemica. Aceasta conditie trebuie sa fie luata in considerare in diagnosticul diferential al cefaleei cu tulburari de vedere la copil.

PSEUDOTUMOR CEREBRI (IDIOPATHIC INTRACRANIAL HYPERTENSION): CAUSE OF HEADACHES AND VISUAL LOSS IN CHILDREN

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Background: Benign intracranial hypertension (BIH) also known as idiopathic intracranial hypertension (IIH) or pseudotumor cerebri, is a neurological disorder that is characterized by increased intracranial pressure, in the absence of a tumor or other diseases affecting the brain. It is a cause of progressive visual loss in children and young adults. The diagnosis of benign intracranial hypertension is mainly clinical, but radiologic and laboratory studies have a role in confirming the diagnosis.
Case report: We report a 6-year-old girl who presented with a 5-day history of headache, visual disturbances (diplopia) and bilateral convergent strabismus. Computer tomography and magnetic resonance of the brain were normal. Ophthalmic examination revealed normal intra-ocular pressure, normal anterior segment and papillary response. The funduscopy revealed papilledema with flu aspect, peripapillary flame hemorrhages, and venous engorgement (chrysanthemum flower). Also she presents limited abduction of the left eye due to the sixth cranial nerve palsy. She was treated with cerebral depletion treatment (mannitol) and corticoids (intravenous dexamethasone and prednisone). The clinical evolution was good, four weeks later there was complete resolution of papilledema.
Conclusion: We present a case of pseudotumor cerebri associated with unilateral sixth cranial nerve palsy, without infectious, toxic, endocrine, metabolic or systemic cause. This condition should be considered in the differential diagnosis of headache with visual loss in children.

NUTRIŢIE ŞI BOLI METABOLICE / NUTRITION AND METABOLIC DISEASE

27. EVALUAREA DISTRIBUTIEI IN POPULATIE A CAZURILOR DE EXCES PONDERAL ŞI OBEZITATE LA COPIL IN JUDETUL MURES

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În conditiile pandemiei de obezitate din ultimii ani,inSUA masurile de sanatate publica au dus la stagnarea cresterii incidentei, inEuropa existand o ingrijorare privind trendul ascendent al acestei tulburari.
Obiectiv:identificarea categoriilor populationale in care prevalenta obezităţii la copil este mai mare, pentru instituirea masurilor profilactico-terapeutice.
Material,metode:s-au efectuat masurători antropometrice în mai multe localitati din Mures la404copii, esantion reprezentativ pentru populatia judetului,considerand o prevalenta a obezitatii de5-20% conform altor studii.Am definit excesul ponderal ca scor-z al Indicelui de masa corporala pentru-vârstă peste1DS, iar obezitate peste2DS.Am analizat distributia cazurilor dupa varsta,sex si mediu de provenienta.

Rezultate:Tulburarile de exces totalizeaza aproape o cincime din copii(17%).Excesul ponderal apare la 1 din14 copii, predominand la10-14ani (global, pe sexe si medii de viata),iar obezitatea la 1 din10 copii.La fetele de 10-14ani, peste un sfert au o tulburare nutritionala de exces,obezitatea predominand la 5-9si 15-19ani;la baieti obezitatea este mai frecventa decat excesul ponderal la toate grupele de varsta,cu pondere maxima la 5-9ani, urmata de10-14 si15-19 ani.Indiferent de mediul de viata,obezitatea predomina la toate grupele de varsta.Excesul ponderal apare in8,33%cazuri in mediul rural fata de6,25% in orase,obezitatea predominand in mediul urban(12,05vs10,55%).
Concluzii:Tulburarile nutritionale de exces la copil înMureş au pondere maximă la10-14 ani,la sexul masculin.Supraponderea este mai frecventă în mediul rural,iar obezitatea in mediul urban.Se impun masuri profilactice, precum ore de educatie pentru sanatate la copiii din clasele primare,promovarea alimentatiei sanatoase si activitatii fizice la toate nivelele,insistand la adolescenti si în orase.
Cuvinte-cheie: copil, exces ponderal, obezitate, sex, varsta

THE ASSESSMENT OF THE DISTRIBUTION IN POPULATION OF CHILD OVERWEIGHT AND OBESITY CASES IN MURES COUNTY

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În condition of obesity pandemy from latest years,inUSA the public health measures leaded to stop in increasing of incidence,in Europe being a concern about the ascendent trend of these disorder.
Objectiv: identification of population cathegories in which the prevalence of obesity is higher,in order to start profilaxy and treatment.
Matherial,methods:we performed anthropometric measurements in several localities inMures,on404children, representative for this conty population,considering the prevalence of obesity 5-20%,according to other studies.Overweight was defined asBodyMassIndex-z-score over1SD,and obesity over2SD.We analysed the cases distribution on age,gender and provenience areas.
Results:Nutritional excess disorders sumarise almost a fifth(17%).Overweight appears in one of 14children, predominant in 10-14years (globally,on sex and provenience area),while obesity in one of 10children.In girls between10-14years, more than a quater have an excess nutritrional disorder,with predominantion of obesity in 5-9and 15-19years;in boys,obesity was more frequent than overweight on all age-groups,with maximum at 5-9years,followed by10-14 and15-19years.Regardless of life environment, obesity prevailed in all age groups.Obesity occurs in 8.33%of cases in rural areas compared to6.25%in cities and obesity prevailed in urban areas(12.05vs10.55%).
Conclusions:nutritional disorders by excess in children inMureş have a maximum on10-14 years age-group,in boys.Overweight is more common in rural areas,while obesity in urban areas.Prophylactic measures are required,such as health education hours in primary school children,promoting healthy eating and physical activity at all levels,focusing on teens and in cities.

28. INTERDEPENDENTA CITOLIZA HEPATICA – SINDROM METABOLIC LA COPIII OBEZI (STUDIU PROSPECTIV)

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Menţiune: Această lucrare este efectuată în cadrul Programului Operaţional Sectorial pentru Dezvoltarea Resurselor Umane (POSDRU), finanţat din Fondul Social European şi Guvernul României prin contractul nr. POSDRU/ 159/1.5/S/137390.

Introducere: Obezitatea reprezinta o problema de sanatate publica, cu o crestere a prevalentei in ultimele decenii aplicabila si populatiei pediatrice. Un rol central in fiziopatologia obezitatii il are afectarea hepatica, implicata in patogeneza sindromului metabolic.
Material, metoda: Studiu prospectiv efectuat in perioada iulie 2014 – martie 2015: copii supraponderali si obezi intre 6 si 18 ani internati in SCUC “G. Alexandrescu” Bucuresti - Pediatrie.
Rezultate, discutii: Au fost inclusi 68 pacienti: 23 supraponderali (IMC > percentila 85) si 45 obezi (IMC > percentila 95), baieti/fete 1.2/1, varsta medie 11.8 ani. Din AHC: 53 de pacienti (77.9%) cu rude apropiate cu obezitate, 48 copii (70.5%) cu cel putin un parinte afectat de obezitate, 14 dintre ei (20.5%) cu ambii parinti obezi. Doar 7.3% dintre copiii au AHC de steatoza hepatica.
Afectare hepatica: 29% (14 copii) prezinta citoliza hepatica si 41% au modificari ecografice (steatoza hepatica).
Sindrom metabolic:19 copii (27.9%) au criterii complete, 18 pacienti (26.4%) au cel putin 2 criterii pozitive pentru acest diagnostic, dintre care unul este constant circumferinta abdominala peste percentila 95.
Concluzii: Sindromul metabolic este o entitate greu de diagnosticat la copii, o proportie semnificativa a pacientilor avand criterii incomplete pentru diagnostic. Afectarea hepatica se asociaza cu sindromul metabolic la copiii din lotul de studiu. Modificarile ecografice (steatoza hepatica) sunt mai frecvente decat citoliza hepatica.
Cuvinte cheie**:** sindrom metabolic, afectare hepatica

PNEUMOLOGIE / PNEUMOLOGY

29. OBTINEREA CONTROLULUI IN ASTMUL ALERGIC SEVER AL COPILULUI PRIN UTILIZAREA DE ANTICORPI MONOCLONALI ANTI- IGE [OMALIZUMAB]

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Astmul sever la copil este o entitate extrem de rara. Excluderea diagnosticelor alternative si obtinerea controlului bolii reprezinta un efort deosebit pentru pneumologul pediatru. Exista relativ putine optiuni terapeutice, una fiind aditionarea de omalizumab [anticorpi monoclonali anti-IgE] la terapia antiinflamatorie inhalatorie si/sau sistemica.

Lucrarea descrie evolutia pe o durata medie de 18 luni a patru scolari [3 baieti si o fata], avand astm alergic documentat.

Anterior includerii aveau astm necontrolat cu ACT<19, in pofida unei bune compliante si a unor doze inalte de corticosteroizi inhalatorii administrati in terapie combinata [cu LABA si/sau LATRA]. Toti intruneau fenotipul exacerbator si aveau asociata rinita/rinoconjunctivita severa si dermatita atopica.

Administrarea s-a facut in prezenta familiei, in cadrul nucleului de resuscitare al CPU IOMC Alfred Rusescu, sub monitorizare continua complexa cu durata variabila 2-4 ore, in regiunea deltoidiana a bratului ales de catre copil, sub analgezie locala cu EMLA.

Nu s-au integistrat efecte adverse severe, singurul element consemnat la toti pacientii fiind febra.

Sunt prezentate graficele evolutive ale valorilor nivelelor de IgE totale, cu aspectul bifazic caracteristic – initial crestere [doi din cei 4 copii au avut cresteri impresionante de IgE totale peste 2000UI/ml] apoi scadere lenta progresiva. Toti pacientii au raspuns favorabil dvdv al controlului astmului, 3 din 4 pacienti finnd partial controlati dar fara exacerbari si unul complet controlat.

CONCLUZII - Omalizumab reprezinta o alternativa terapeutica sigura si eficace la corticosteroizii sistemici pentru pacientii de varsta pediatrica [varsta peste 6 ani] cu astm alergic sever necontrolat.

CUVINTE CHEIE – astm, alergie, omalizumab, copil

CONTROLLING SEVERE ALLERGIC ASTHMA WITH OMALIZUMAB (MONOCLONAL ANTI-IGE ANTIBODIES) IN CHILDREN

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Allergic severe asthma is a relative rare disease among young children. Most of these patients should be evaluated by an expert and treatment has to be tailored according to evolutive phenotype. One option for adult patients with such a condition is adding omalizumab [anti-IgE antibodies] to step IV GINA medication.

Authors present a small pediatric series of severe uncontrolled asthmatic patients treated with an average 18 months course of omalizumab. Demographics, clinical features and comorbid conditions are documented. All patients were treated in the resuscitation module of the Emergency Department from a tertiary referral pediatric hospital. Monitoring and protocol are presented. All children had a positive outcome, three with partial control and one with complete control. No serious side effects were observed. Low grade fever was easily controlled with trivial antipyretics.

Conclusion – monoclonal antibodies against IgE in severe allergic asthma patients are efficacious also in children and represent a safe and solid alternative for long-term oral corticosteroid treatment.

KEY WORDS – allergic asthma, anti IgE antibodies, child

30. ROLUL OXIDULUI NITRIC EXPIRAT IN OBIECTIVAREA DISFUNCTIEI OBSTRUCTIVE DIN ASTMUL BRONSIC

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Introducere. Valoarea fractiei oxidului nitric expirat (FENO) detine statutul de biomarker noninvaziv in astmul bronsic. Rolul investigatiei in stabilirea diagnosticului de astm bronsic este inca in curs de definire. Scopul studiului a fost investigarea dinamicii relatiei dintre nivelul FENO si valorile parametrilor spirometrici specifici obstructiei bronsice.

Materiale şi metode. Studiul longitudinal a inclus un lot de 89 de copii preluati din cazuistica Clinicii de Pediatrie. Criteriile de includere au fost: (i) diagnosticul prestabilit de astm bronsic, (ii) varsta cuprinsa intre 5 si 18 ani. Tuturor pacientilor li s-au efectuat probe functionale respiratorii si li s-a determinat valoarea oxidului nitric expirat.

Rezultate. Valorile FENO s-au corelat semnificativ statistic semnificativ doar cu o parte dintre parametrii spirometrici: MEF75 (p=0.007), MEF50 (p=0.0.28), MEF25 (p=0.011) si PEF (p=0.020), dar nu si cu FEV1 sau FVC. Regresia liniara multipla a relevat ca variatia FENO nu poate fi estimata decat intr-un grad redus de catre parametrii spirometrici (R=0.062, p=0.82). Iar regresia logistica binara a aratat ca FENO nu este un biomarker cu semnificatie statistica (p=0.169) pentru estimarea riscului de disfunctie obstructiva.

Concluzii

Nivelul oxidului nitric expirat nu poate fi utilizat pentru obiectivarea disfunctiei obstructive din astmul bronsic.

Cuvinte cheie

FENO, astm bronsic, disfunctie obstructiva

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31. CAUZA RARA DE INSUFICIENTA RESPIRATORIE ACUTA LA NOU NASCUT

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Prezentam cazul unui nou-nascut în varsta de 15 zile internat in Clinica de Pediatrie cu semne de insuficienta respiratorie acuta.

Nou nascutul, din sarcina dispensarizata, greutate la nastere 3440g, Apgar 9, a prezentat cu 2-3 zile anterior internarii, oboseala la alimentatie, dificultati de respiratie.

La internare: afebril, 3250g, cu cianoza perioronazala accentuata la plans, dispnee mixta cu polipnee, FR 70/min, fara tuse, balans toraco-abdominal, saturatii O2 90%, hernie ombilicala, echilibrat hemodinamic si digestiv.

Paraclinic: HLG normala, fara sindrom biologic inflamator, acidoza respiratorie cu hipercapnie. Radiografia pulmonara a evidentiat imagini mixte de opacitati si hipertransparenta (accentuate in LSS) cu deplasarea mediastinului spre stanga, hipertransparenta hemitoracelui drept, diafragme aplatizate. S-a suspectat hernie diafragmatica, infirmata de tranzitul baritat.

In evolutie, la 2 zile a prezentat accentuarea dispneei, saturatii O2<90% cu pneumotorax drept drenat cu expansionare partiala a plamanului drept. S-a efectuat CT torace si s-a ridicat suspiciunea de malformatie pulmonara. In serviciul de ATI prezinta recidiva de pneumotorax drept, drenat. Se transfera de urgenta in serviciul de terapie intensiva neonatala, Sp Marie Curie Bucuresti.

S-a intervenit chirurgical: lobectomie lob superior drept, evidentiindu-se malformatie adenomatoida chistica congenitala dreapta, cu evolutie postoperatorie favorabila.

Discutii: malformatia chistica adenomatoida congenitala este rara, reprezentand o anomalie de dezvoltare pulmonara, ce poate fi descoperita intamplator.

Cazul prezentat a debutat cu insuficienta respiratorie acuta, complicata cu pneumotorax spontan, ceea ce putea avea prognostic nefavorabil.

Este necesara recunoasterea precoce a malformatiilor pulmonare, mai ales in context de manifestari clinice acute, in vederea rezolvarii optime chirurgicale si prevenirii complicatiilor.

A RARE CAUSE OF RESPIRATORY DISTRESS IN A NEONATE

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We report a case of a 15 days old male neonate, admitted to the Pediatric Clinic with respiratory distress.

He was born at 39 weeks of gestation, 3440g, Apgar score 9 and had no significant medical history before hospitalization.

2 days before admittance he presented exertion at feeding, difficult breathing.

Physical examination revealed an afebrile, normal weighted infant with respiratory distress, tachypnoea, oxygen saturation of 90% , perioral cyanosis during crying, no cough. Cardiovascular and digestive examination were normal. CBC and CRP were normal; there was respiratory acidosis, hypercapnia. The chest X-ray revealed mixed diffuse opacities of the LUL, hypertransparency in the LUL and right lung field, left shift of the medistinum, flattened diaphragm. Barium swallow test was negative for diaphragmatic hernia.

The 2nd day of hospitalisation dyspnea exacerbated, oxygen saturation decreased. Right pneumothorax developed, managed by tube-drainage, with partial expansion of the lung. Chest CT-scan showed suggestive images for pulmonary malformation. Few days later, a second pneumothorax occurred, which was drained. He was reffered to the Emergency Clinical Hospital for Children Marie Curie, Bucharest and underwent right upper lobectomy with favorable outcome. Histologic examination confirmed CCAM.

Discutions: cystic adenomatoid malformation is a rare congenital abnormality of lung development, may present as an incidental finding.
The reported patient presented with respiratory distress followed by recurrent spontaneous pneumothorax. Prognosis could have been poor. Pulmonary malformation should be considered, especially in case of acute symptoms, in order to corectly manage and prevent any further complications.

32. ASPECTE EVOLUTIVE IN FIBROZA CHISTICA- O BOALA CU MAI MULTE FETE

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Introducere: Tabloul clasic al fibrozei chistice(mucoviscidoza) include, pe langa pneumopatia cronica si steeatoree , o multitudine de alte manifestari ca hepatopatia, diabetul, deficitul nutritional, care pot constitui modalitate de debut a bolii. Polimorfismul manifestarilor dar si evolutia diferita a pacientilor cu o maladie monogenica ca si fibroza chistica(FC) ridica probleme de diagnostic si monitorizare in practica zilnica. Scopul lucrarii este de a supune atentiei cateva modalitati de debut si evolutie a pacientilor cu FC.
Metode: Inca de la descoperirea genei FC si a mutatiilor genetice s-a vehiculat conceptul influentei fenotipului de catre genotip, dar evolutia diferita a pacientilor cu acelasi genotip a infirmat , de-a lungul timpului,aceasta corelatie. Exceptand pneumopatia si insuficienta pancreatica exocrina, ciroza hepatica sau osteopatia pot fi modalitati de manifestare a FC. Rezultate: Lucrarea prezinta evolutii diverse ale pacientilor cu genotip comun si modalitati de debut mai mult sau mai putin particulare. Concluzii: Individualitatea se manifesta pregnant si la pacientii cu FC, nu exista „boala" ci doar pacient. Este din ce in ce mai probabil ca factori diferiti de genotip si mediu sa influenteze evolutia si prognosticul copiilor cu fibroza chistica.

OUTCOME IN CYSTIC FIBROSIS-A DISEASE WITH MANY FACES

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Background: The classic picture of cystic fibrosis (CF) includes, in addition to chronic lung disease and steeathoreea, a multitude of other clinical features like: CF associated liver disease, CF related diabetes, nutritional deficiencies, which can be way onset of the disease. Polymorphism and different outcome of patients with same genotype, in a monogenic disease as CF raises questions of diagnosis and monitoring in daily practice. The purpose of this paper is to present few ways of diseases onset and evolution among CF patients.
Methods: Since the CF gene discovery the concept of genotype/ phenotype correlation evolved, but different evolution of patients with the same genotype denied, over time, this correlation. Except pneumonitis and exocrine pancreatic insufficiency, cirrhosis or osteopathy may be ways of CF onset. Results: This paper presents the evolution of patients with identic genotype and various types of onset and more or less particular. Conclusions: Individuality is strongly manifested in patients with CF, there isn't only "disease" but patient. It is increasingly likely that other factors, than genotype and environment, influence the evolution and prognosis of children with cystic fibrosis.

33. INFECTII SEVERE DE TRACT RESPIRATOR INFERIOR LA SUGAR SI COPILUL MIC- STUDIU COMPARATIV PE 4 ANI

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Infecţiile de tract respirator inferior (ITRI) reprezintă o cauză importantă de morbiditate şi mortalitate
OBIECTIVE: Compararea ITRI de etiologie virala cu ITRI de etiologie bacteriană la copii.
MATERIAL SI METODA: Studiu retrospectiv pe 4 ani, au fost selectaţi copii între 0-5 ani cu infecţie severă de tract respirator inferior.
REZULTATE: 72 de copii au avut stabilit un diagnostic etiologic concret de infecţie virala. Etiologia a fost reprezentată de virusul sinciţial respirator 73,5%, Influenza 11%, Parainfluenza 4,2%, Metapneumovirusul, Rinovirus, Bocavirus şi Adenovirus toate cu 2.8%. Etiologia bacteriană a putut fi stabilită in 30 % din cazuri, Streptoccocus Pneumoniae fiind cel mai frecvent implicat. În cadrul infecţiei virale rata de folosire a antibioticelor a fost de 92%. În cadrul infecţiei bacteriene antibioticele au fost folosite în toate cazurile. Durata spitalizării a fost în medie cu 2 zile mai mare în cadrul lotului cu infecţie virală specificată faţa de cei cu infecţie virală nespecificată.
CONCLUZII: Rata de diagnostic etiologic în cadrul infecţiilor virale este încă foarte redusă. Antibioterapia este utilizată frecvent în formele severe de ITRI, chiar dacă etiologia virală este certă şi cunoscută. Determinarea etiologiei virale nu a redus spitalizarea, şi nici nu a dus la schimbarea atitudinii terapeutice a medicului curant.

Menţiune : Această lucrare este efectuată în cadrul Programului Operaţional Sectorial pentru Dezvoltarea Resurselor Umane (POSDRU), finanţat din Fondul Social European şi Guvernul României prin contractul nr: POSDRU/159/1.5/S/137390

SEVERE LOWER RESPIRATORY TRACT INFECTIONS IN INFANTS AND CHILDREN - 4 YEARS COMPARATIVE STUDY

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Lower respiratory tract infections represent an important cause of morbidity and mortality in pediatric pathology

OBJECTIVES: Comparison of severe lower respiratory tract infections (LRTI) of viral etiology with LTRI of bacterial etiology in children

MATERIAL AND METHODS: Retrospective study over a period of 4 years, were selected children 0-5 years with severe lower respiratory tract infection.

RESULTS: 72 children who have shown signs of acute respiratory failure have been diagnosed with a specific viral infection. The main etiology was represented by respiratory syncytial virus (RSV) in 73.5% of cases, followed by Influenza 11%, Parainfluenza 4.2%, Metapneumovirus (hMPV), Rhinovirus, Adenovirus and Bocavirus all with 2.8%. The bacterial etiology was determined in 30% of cases, Streptoccocus pneumoniae was the most commonly involved. In the context of viral infection, antibiotics were used in 92% of cases. For the bacterial infection antibiotics were used in all cases. The length of hospital stay was 2 days higher in the group with specified viral infection compared with the group with unspecified viral infection.

CONCLUSIONS: In severe LRTI of viral etiology the etiological diagnosis is rarely established. Antibiotics are commonly used in the severe forms of LTRI, even if the viral etiology is certain. Determination of viral etiology has not reduced hospitalization, nor has led to changing of physician therapeutic approach

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REUMATOLOGIE / RHEUMATOLOGY

34. AIJ: LUNGUL DRUM SPRE CASA

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Artrita idiopatica juvenila (AIJ) reprezinta un capitol important din patologia cronica pediatrica, avand o incidenta de ordinul 10/100.000 de persoane pe an dar un impact personal, familial si socioeconomic mult mai mare. Deoarece nu exista un test unic cu rol de standard de aur, diagnosticul se bazeaza pe seturi de criterii, majoritatea clinice. Testele paraclinice si de laborator au rol doar in excluderea diagnosticelor alternative, ceea ce, daca este facut cu rigurozitate, asigura un bun diagnostic pozitiv. Erorile in acest sens sunt bidirectionale si sunt des intalnite in cazuistica noastra. Astfel, multi dintre pacientii cu AIJ pauciarticulara au fost diagnosticati la debut ca artrite septice sau entorse si tratati cu metode ortopedice locale (imobilizare, rivanol rece) sau sistemice (antibiotice). La celelalt capat al spectrului se aflau diverse boli diagnosticate (si tratate initial) ca AIJ: mucocel maxilar odontogen, lupus eritematos sistemic (LES), limfom Hodgkin, limfohistiocitoza hemofagocitica, sarcom iliac, artrita vilonodulara, corp strain. In procesul diagnostic al AIJ, trebuie explorata fiecare posibila explicatie alternativa, deoarece tratamentul si prognosticul pot fi total diferite, atat in bine, cat si in rau.

JIA: THE LONG WAY HOME

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Juvenile idiopathic arthritis (JIA) is a chronic pediatric pathology important chapter, with an incidence of 10/100000 people per year but a much higher personal, familial, social and economic impact. Since no gold standard diagnostic test exists, diagnosis is based on sets of criteria, most of them clinical. Other tests only serve to exclude alternative diagnoses, which, if done rigorously, ensures proper positive diagnosis. Errors in this respect are bidirectional and frequently encountered in our

experience. Thus, many patients with pauciarticular JIA were initially diagnosed as septic arthritis or sprains and were treated with local orthopedic (immobilization, cold antiseptic solutions) or systemic methods (antibiotics). At the other end of the spectrum were various diseases, diagnosed (and originally treated) as JIA: maxillar odontogenic cyst, systemic lupus erythematosus (SLE), Hodgkin's disease, hemophagocytic lymphohistiocytosis, pelvic sarcoma, villonodular arthritis, foreign articular body. In JIA diagnosing process, every possible alternative explanation must be explored, as treatment and prognosis may be very different, either for the better of for the worse.

Cuvinte cheie: artrita idiopatica juvenila, diagnostic diferential
Keywords: juvenile idiopathic arthritis, differentials

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35. DUREREA MUSCULOSCHELETALA LA COPIL: DE LA SIMPTOM LA DIAGNOSTIC COMPLEX

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Introducere

Durerea musculoscheletală reprezintă suferinţa algică localizată la oricare dintre structurile sau elementele componente ale aparatului locomotor, de cauza posttraumatica, inflamatorie, infecţioasa, degenerativa sau maligna. Diagnosticul diferenţial al durerii musculoscheletale trebuie realizat atent si complet.

Obiective

Autorii investighează durerea musculoscheletală de origine reumatică (artrită juvenilă idiopatică, lupus eritematos sistemic, purpură Henoch-Schonlein, dermatomiozită, artrită psoriazică, artrită reactivă), de origine non-reumatică dar asociată cu modificări organice (hipermobilitate, obezitate, alte patologii articulare, osoase) și durerea din alte afecțiuni (alergii, durere idiopatică).

Material și metode

Lotul de studiu a fost constituit din 184 copii internați în Clinica II Pediatrie Iași în cursul anului 2014 pentru durere articulară sau musculară. S-a studiat aportul termografiei cutanate și al electromiografiei în investigarea acestor pacienți.

Rezultate și discuții

Durerea articulară sau musculară a reprezentat 18,2% din totalul internărilor în Clinica II Pediatrie Iași în anul 2014. Profilul diagnostic a relevat artrita juvenilă idiopatică, lupusul eritematos sistemic, purpura Henoch-Schonlein, spasmofilia, obezitatea, dermatomiozită / polimiozită, miopatii congenitale, artrită psoriazică, artrită reactivă. Dizabilitatea funcțională se corelează cu intensitatea durerii și este însoțită de modificări psiho-afective. Electromiograma efectuată la 82 pacienți a relevat trasee miogene în 35 cazuri (42,68%) și neurogene în 10 cazuri (12,2%). Termografia cutanată a evidențiat modificările de temperatură la nivelul diferitelor segmente ale corpului, corelate cu sediul și intensitatea durerii.

Concluzii

Durerea musculoscheletală la copil necesită diagnostic diferențial atent din partea clinicianului. Electromiografia și termografia cutanată sunt metode complementare ce completează tabloul investigațiilor paraclinice în durerea musculoscheletală la copil.

MUSCULOSKELETAL PAIN IN CHILDREN: FROM SYMPTOMS TO COMPLEX DIAGNOSIS

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Introduction

Musculoskeletal pain is localized in any structure or component of musculoskeletal system, often posttraumatic, inflammatory, infectious, degenerative or malignant. Differential diagnosis of musculoskeletal pain should be done carefully and completely.

Objectives

Authors investigate musculoskeletal pain of rheumatic origin (juvenile idiopathic arthritis, systemic lupus erythematosus, Henoch-Schonlein purpura, dermatomyositis, psoriatic arthritis, reactive arthritis), non-rheumatic origin but associated with organic disorders (hypermobility, obesity and other joint or bone diseases) and pain from other conditions (allergies, idiopathic pain).

Material and methods

The study group consisted of 184 children hospitalized in 2nd Pediatrics Clinic of Iasi during 2014 admitted for joint or muscle pain. It is shown the contribution of skin thermography and electromyography in investigating those patients.

Results and discussion

Joint or muscle pain represented 18.2% of total admissions in 2nd Pediatrics Clinic of Iasi in 2014. Complete diagnosis revealed juvenile idiopathic arthritis, systemic lupus erythematosus, Henoch-Schonlein, spasmophilia, obesity, dermatomyositis / polymyositis, congenital myopathies, psoriatic arthritis, reactive arthritis. Correlations with functional disability and pain intensity were made along with psycho-emotional changes. Electromyogram performed in 82 patients

showed myogenic pattern in 35 cases (42.68%) and neurogenic pattern in 10 cases (12.2%). Thermography revealed skin temperature changes in various parts of the body, correlated with pain intensity.
Conclusion
Musculoskeletal pain in children requires careful differential diagnosis by the clinician. Electromyography and skin thermography are complementary methods of investigations completing the diagnosis in musculoskeletal pain in children.

36. VARIANTELE EVOLUTIVE ALE ARTRITEI JUVENILE IDIOPATICE IN BAZA REGISTRULUI NATIONAL AL MALADIILOR REUMATICE LA COPII

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Introducere. Artrita idiopatica juvenila (AIJ) - cea mai intalnita forma de artrita inflamatorie la copii cu 7 variante evolutive constituie una dintre cele mai severe boli cronice reumatologice in grupa de varsta 1-16 ani.
Scopul. Determinarea ponderii variantelor evolutive a AJI in Registrul National al maladiilor reumatice la copii.
Materiale si metode. In perioada anilor 2012-2014 au fost chestionati 338 copii cu diverse maladii reumatice. Rezultate. In baza registrului National al maladiilor reumatice AJI a constituit 27% (91 pacienti), dintre care: 44 % copii au fost cu oligoartrita, 32% cu poliartrita, 15% cu varianta sistemica, 8 % - AJI in asociere cu entezite si doar la 1% - artrita psoriazica. La momentul examinarii manifestau artrita activa 94,5 % pacienti. La copii cu varianta oligo- si poliarticulara predomina afectarea genunchilor (70% vs 76%), iar in varianta sistemica s-a notat afectarea și articulatiilor pumnului (78%). De notat ca 59% copii cu varianta oligoarticulara in activitatea cotidiana manifestau dificultăti usoare, atunci cind copiii cu varianta poliarticulara si sistemica au relatat dificultăti severe. Circa 50 % pacienti cu AJI au o stare functionala deficitara cu divers grad de invaliditate.
Concluzii. In structura AJI variantele poliarticulare si sistemice sunt cu impact major asupra functiei articulare si calitatii vietii copiilor.

EVOLUTIVE ALTERNATIVES OF JUVENILE IDIOPATHIC ARTHRITIS BASED ON THE NATIONAL REGISTER OF RHEUMATIC DISEASES OF CHILDREN

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Introduction. Juvenile Idiopathic Arthritis (JIA) - the most common form of inflammatory arthritis in children, with 7 evolutional alternatives is one of the most severe chronic rheumatologic diseases in the age group 1-16 years.
The Goal. Determining the share of the evolutive alternatives of JIA in the National Register of Rheumatic Diseases of children.
Material and methods. During the years 2012-2014, 338 children were examined with various rheumatic diseases.
Results. Based on the National Register of Rheumatic Diseases, JIA counted 27% (91 patients), where 44% - children with oligoarthritis, 32% - with polyarthritis, 15% - with systemic variant, 8% - JIA in association with enthesitis and only 1% - with psoriatic arthritis. During the examination 94.5% of the patients expressed active arthritis. To the children with oligo- and poliarticular version, there was knee damage noticed (70% vs 76%), and in the systemic variant was also noticed the damage of the wrist (78%). Is worth mentioning that 59% of children with oligoarticular version showed minor difficulties in the everyday life, while the children with polyarticular and systemic variants reported severe difficulties. About 50% of the patients with JIA have a poor functional condition with different degrees of disability.
Conclusions. In the JIA structure, polyarticular and systemic variants are having a major impact on the joint function and quality of life of children.

URGENŢE – TOXICOLOGIE / PEDIATRIC EMERGENCIAS – CLINIC TOXICOLOGY

37. PARTICULARITATI PEDIATRICE IN TEHNICA DE CONTROL AVANSAT A CAII AERIENE

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Evaluarea și luarea unor decizii corecte și rapide pot preveni evoluția către insuficientă respiratorie, stop respirator (SR) sau stop cardiorespirator (SCR). Spre deosebire de adulții aflați într-o situație amenințătoare de viață unde se înregistrează mai frecvent stop cardiac ca urmare a deteriorării primare a statusului circulator (aritmii cu ritm de colaps: fibrilație ventriculară, tahicardie ventriculară fără puls, asistolie, disociație electromecanică), la copii este consemnată apariția stopului cardiac în urma deteriorării progresive a funcției respiratorii, ca urmare a unor afecțiuni pulmonare sau extra-pulmonare severe. Dacă stopul respirator este prelungit, producând și asistolie (SCR) șansele de supraviețuire scad de 10 ori (7 – 11% supraviețuire în SCR fată de 70 – 90% in SR).

La copilul aflat într-o situație extremă ar trebui anticipată deteriorarea iar măsurile ar trebui inițiate cat mai precoce. Abordarea tardivă a copilului cu stop respirator scade drastic șansele de supraviețuire și creste riscul de sechele neurologice.

Din rațiuni etice manevrele de învățare ale controlului avansat al căii aeriene la copil nu pot fi făcute în cadrul unor manevre reale de resuscitare. Pentru a realiza o stăpânire eficientă a acestor manevre la copil [poziționarea caii aeriene, deschiderea acesteia și menținerea ei cu diverse dispozitive (pipa Guedel, pipa nazo-faringiana), plasarea sondei nazo-gastrice, ventilația cu balon și masca, plasarea măștii laringiene sau intubația traheală] este necesară repetarea acestora în scenarii clinice, în cadrul unor ateliere de simulare.

Sunt descrise atelierele folosite in cadrul cursului PALS [Pediatric Advanced Life Support] din IOMC și este documentată creșterea eficientei clinice prin repetarea constantă și periodică a manevrelor.

CUVINTE CHEIE – simulare, resuscitare, cale aeriană, copil

PEDIATRIC ASPECTS IN ADVANCED AIRWAY CONTROL

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Rapid assessment of airway and timely action can prevent deterioration of respiratory failure to respiratory arrest. In adults arrest can be frequently generated by a significant cardiac condition with a collapse rhythm [ventricular fibrillation, ventricular tachycardia without pulse, asystole and pulseless electrical activity] as opposed to children that exhibit more often hypoxemic arrest due to progressive worsening of a condition that impairs gas exchange.

If such an arrest situation is prolonged to such an extent to progress from respiratory arrest to cardio-respiratory arrest survival is 10 times less likely and a decrease of resuscitation success from 70-90% to 7-11% can be documented.

Due to ethical reasons advanced airway control cannot be taught or exercised during real-life resuscitation in children. In order to achieve proficiency in airway management in children simulation with adequate mannequins can be used. Various scenarios can be imagined for airway positioning, bag and mask ventilation, laryngeal mask insertion or tracheal intubation.

Authors describe strategies used during PALS course in order to simulate airway control and provide solid proof that repetitive training can achieve proficiency in this area of pediatric resuscitation.

KEY WORDS – simulation, resuscitation, airway, child

38. INTOXICATIILE ACUTE SEVERE LA COPII – STUDIU RETROSPECTIV PE O PERIOADA DE 5 ANI

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Obiective: analiza etiologiei intoxicatiilor acute la copii, complicate cu soc si evolutie spre deces. Material si metoda: am analizat toate cazurile de intoxicatii acute, pe o perioada de 5 ani, care s-au complicat cu soc si deces. Criteriile de soc: tahicardie, tahipnee, prelungirea timpului de recolorare capilara, hipotensiune arteriala si agitatie. Au fost selectate 78 de cazuri. Toti pacientii au fost internati in Terapie Intensiva, unde s-a instituit rapid tratamentul suportiv si antidotic. Rezultate: Etiologia intoxicatiilor acute complicate cu soc a fost urmatoarea: 57 cazuri de substante de abuz /medicamente (12 substante de abuz: 3 heroina, 3 canabis si 6 substante cu actiune opioid-like, 13 nafazolina, 8 Dentocalmin - lidocaina, 8 cazuri de betablocante – Distonocalm, propranolol, metoprolol, 6 cazuri de ADTC – amitriptilina, imipramina, 3 cazuri clonidina, 2 digoxin si cate un caz: fenobarbital, bromazepam, salbutamol, isosorbid dinitrat, miofilin retard – teofilin), 6 cazuri de insecticide (5 organofosforice – diazinon si 1 carbamat – furadan), 7 cazuri de nitriti, 4 alcool si un caz de intoxicatie cu monoxid de carbon. De asemenea, 3 cazuri de intoxicatie cu ciuperci au asociat soc hipovolemic. Din totalul de 78 de cazuri,

13 pacienti au decedat (etiologie: 7 droguri, 6 Dentocalmin, 1 amitriptilina, 5 pesticide si un caz de nitriti) iar 2 cazuri au dezvoltat ulterior complicatii neurologice severe. Concluzii: instituirea rapida a tratamentului antidotic si suportiv a dus la recuperarea completa a starii de sanatate a copiilor. Intoxicatiile acute la varsta pediatrica inca reprezinta o problema e sanatate publica in tara noastra, unele dintre acestea evoluand cu complicatii severe inclusiv deces. Cuvinte cheie: intoxicatie, soc, deces.

SEVERE ACUTE POISONING IN CHILDREN – A 5-YEAR RETROSPECTIVE STUDY

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Objective: analisys of the etiology of acute poisoning in children complicated with shock and death.
Materials and method: we analysed all cases of acute poisoning in a 5 year period that where complicated with shock and death. Shock criteria: tahicardia, tahipnea, prolongated capilary refill time, low blood pressure and iritability. There were selected 78 cases. All patients were admitted in ICU and supportive and antidotic treatment were rapidly started.
Results: Etiology of acute poisoning complicated with shock was: drugs 57 cases (12 cases abuse substances: 3 heroine, 3 cannabis and 6 opioid acting like substances, 13 cases naphasoline, 8 cases Dentocalmin (lidocaine), 8 cases betablockers - Distonocalm, propranolol, metoprolol, 6 cases TCAD - amitriptilin, imipramin, 3 cases – clonidine, 2 cases digoxin, 1 case – fenobarbital, bromazepam, salbutamol, isosorbid dinitrat, miofilin retard - teophylline), 6 cases insecticides (5 organofosforates – diazinon and 1 carbamate – furadan), 7 cases – nitrites, 4 cases – alcohol and 1 case carbon monoxide. Also 3 cases with mushroom poisoning were clasified as hipovolemic shock.
In the total of 78 cases 13 patients died (etiology: drugs 7 cases – 6 cases Dentocalmin and 1 case amitriptilin, pesticides 5 cases, nitrites 1 case) and 2 cases developed severe neurological complications.
Conclusion: due to the rapid unset of supportive and antidotic treatment most of the children recovered to full health status. Acute poisoning in children is still a public health problem in our country, some of them with severe complications in evolution and even death.
Key words: poisoning, shock, death.

TINERI CERCETATORI - REZIDENTI / YOUNG RESEARCHS

39. CAPCANE DIAGNOSTICE IN EXOFTALMIA UNILATERALA LA COPII

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Glaucomul congenital este cea mai frecventa cauza de buftalmie la copii. Buftalmia impune diferentierea de exoftalmie, avand in vedere spectrul etiologic si abordarea terapeutica complet diferite. Exoftalmia unilaterala la copil este rara, putand avea etiologie traumatica, inflamatorie, vasculara, parazitara sau tumorala. Se prezinta doi pacienti in varsta de 9 luni si respectiv 14 ani care au dezvoltat exoftalmie unilaterala rapid progresiva in decurs de 3 saptamani. Examenul oftalmologic a exclus glaucomul, evidentiand glob ocular protruzionat. Evaluarea imagistica a decelat la ambele cazuri prezenta unei tumori de sinus maxilar. Analiza imunohistochimica a permis formularea diagnosticului de sarcom asociat unei leucemii acute monocitare la sugar si respectiv de rabdomiosarcom embrionar de sinus maxilar stang la pacientul de 14 ani. Pacientii au fost tratati prin chimioterapie agresiva cu raspuns initial favorabil. Exoftalmia unilaterala la copil poate fi primul semn al unei patologii tumorale maligne care sa necesite o terapie precoce si sustinuta.
Cuvinte cheie: exoftalmie, leucemie acuta monocitara, rabdomiosarcom embrionar, copii.

DIAGNOSTIC DILEMMA IN RAPIDLY PROGRESSING UNILATERAL EXOPHTHALMIA IN CHILDREN

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Congenital glaucoma is the most common cause of buphtalmos in children. Buphtalmos must be first differentiated from exophthalmia because of the completely different etiologic and therapeutic approach. Unilateral exophthalmia in children is rare, having a traumatic, inflammatory, vascular, parasitic or tumor etiology. We present an infant of 9 months and a teenager of 14 years who both developed a unilateral important exophthalmia within three weeks. The ophthalmological exam excluded glaucoma, finding a unilateral protrusion of the eyeball. The imagistic exams documented in both cases an extended maxillary sinus tumor. The immunohistochemical examination allowed the diagnosis of an acute monocytic leukemia associated with sarcoma in the infant and of an embryonal rhabdomyosarcoma of the left maxillary sinus in the teenager. Both patients were treated with a very aggressive chemotherapy with an initially favorable response. Rapidly progressing unilateral exophthalmia in children may be the first sign of a malignant tumor that requires an early and sustained therapy.
Keywords: exophthalmia, acute monocytic leukemia, embryonal rhabdomyosarcoma, children.

40. CAUZE RARE DE WHEEZING RECURENT- NU ORICE WHEEZING ESTE ASTM!

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Introducere: Wheezing-ul recurent este o patologie frecvent intalnita in practica pediatrica zilnica. De la astm bronsic si tuberculoza pana la boli rare ca deficit de alfa 1 antitripsina, bronsiectazii, etiologia poate fi variata , iar diagnosticul uneori extrem de dificil. Material si metoda: Lucrarea de fata isi propune sa prezinte doua cazuri dewheezing recurent, cazul unei fetite de 10 luni, cu istoric indelungat de 2 luni , referita clinicii cu atelectaziede lob superior drept. La internare se constata stare generala buna, wheezing neresponsiv la betamimetic, radiologic o hiperaeratie evidenta la nivel pulmonar stang. Pe parcursul evolutiei se constata persistenta wheezing-ului inconstant insotit de un sunet de transmisie, suspectandu-se un proces compresiv extern. Totodata se constata ecografic nevizualizarea emergentei trunchiului coronarelor; coroborandu-se datele se recomanda angio CT , ce releva artera lusoria stanga.Un alt caz indrumat clinicii cu suspiciunea de astm bronsic, prezinta, in contextul unei hipotrofii ponderale, tuse cronica si infectie cu Pseudomonas aeruginosa; testul sudorii efectuat arata valori net pozitive ale Cl, in contextul unui genotip heterozigot compus. Concluzie: Nu orice caz de wheezing este astm, chiar daca exista suspiciunea unei patologii alergice; cauza mai rare de wheezing trebuie luate in considerare atunci cand treneaza , este neresponsiv la betamimetic si asociaza si alte complicatii sugestive.

RARE CAUSE OF RECURRENT WHEEZE

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Introduction: Recurrent wheezing is a pathology frequently observed in daily pediatric practice. From asthma and tuberculosis to rare diseases like alpha 1 antitrypsin deficiency, bronchiectasis, it can be varied etiology and diagnosis extremely difficult sometimes. Methods: This paper aims to present two cases with recurrent wheezing :the case of a girl of 10 months, with 2 months long history, referenced for right upper lobe atelectasis. On admission, finds good general condition, wheezing unresponsive to bronchodilators, with radiologic evidence of left lung hyperinflation. During evolution wheezing persisted, accompanied by a strange transmission sound. Also cardiac ultrasound could not revealed the emergence of coronary trunk; Corroborating data a vascular compression was suspected and CT angiography confirmed the case of artery lusoria . Another 2 years old child suspected for asthma, shows, in the context of a hypotrophy weight, chronic cough and infection with Pseudomonas aeruginosa; sweat test was positive, in the context of a compound heterozygous genotype. Conclusion: Not all wheezing is asthma case, even if there is suspicion of allergic diseases; rare cause of wheezing should be considered when chronic evolution, nonresponsive to treatment is present and other complications associated.

41. DIAGNOSTICUL VARSATURILOR LA PREMATUR - O ADEVARATA PROVOCARE

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Varsaturile sunt unul dintre cele mai frecvente simptome in pediatrie, cu o pondere de 50% la sugari din care doar 5% au de obicei o cauza organica, restul fiind simptome fucnctionale, date de imaturitatea tractului digestiv. La prematuri, tractul

digestiv prezinta o malfunctie mai exprimata, motiv care face ca varsaturile sa fie mai frecvente. Acestea afecteaza cresterea si dezvoltarea, problema care capata o importanta deosebita atunci cand vorbim de prematuri de grad mare.
Va prezint cazul unui sugar in varsta de 7 luni, nascut la 26/27 saptamani de gestatie cu o greutate de 660 grame, cu detresa respiratorie severa, hemoragie intraventriculara bilaterala gradul II/III, defect de sept atrial de tip ostium secundum, canal arterial persistent, hernie ombilicala, hemangiom fronto-parietal drept, retinopatie de prematuritate stadiul II. Se prezinta in clinica pentru varsaturi, inapetenta si stagnare ponderala; pana la varsta de 6 luni a prezentat o dezvoltare satisfacatare cu atingerea greutatiii de 4050 grame.
Cazul a reprezentat o adevarata provocare diagnostica. Diagnosticul diferentiala s-a facut intre gastrita acuta, boala de reflux gastro-esofagian, intoleranta la proteinele laptelui de vaca, boala celiaca, fibroza chistica, lactobezoar, malformatii digestive si in special stenoza hipertrofica de pilor, infectie digestiva sau extradigestiva.
Stabilirea diagnosticului pozitiv a fost deosebit de importanta datorita afectarii cresterii la un prematur de grad mare.
In cursul prezentarii voi explica demersul diagnostic efectuat pentru a exclude variantele plauzibile de diagnostic diferential si explorarile efectuate pentru a stabili diagnosticul pozitiv. ; voi discuta dificultatile diagnostice intalnite in cazul varsaturilor prezente la pacientul pediatric prematur.

THE CHALLENGE IN PREMATURE BABY'S VOMITING

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42. CAUZA RARA DE INSUFICIENTA RESPIRATORIE ACUTA LA UN COPIL CU HERNIE HEPATICA DIAFRAGMATICA

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Hernia diafragmatica congenitala (HDC) este una dintre cele mai comune malformatii la nou-nascut si apare la 1 din 2500-5000 de nasteri. Cel mai frecvent tip de HDC este reprezentat printr-un defect posterolateral (foramen Bochdalek) si este responsabil pentru aproximativ 85% din cazuri.
HDC dreapta (foramen Morgagni) este mai putin intalnita (3%) si este asimptomatica la copii, find responsabila de intarzierea in stabilirea diagnosticului. Hernierea ficatului prin foramen Morgagni poate exercita compresiune asupra organelor intratoracice ipsilaerale. Spectrul de prezentare variaza de la insuficienta respiratorie acuta de la nastere, pana la simptome minime sau absente ce pot aparea mult mai tarziu, in cursul vietii, si reprezinta o provocare de diagnostic.
Am raportat un caz de hernie hepatica diafragmatica, descoperita la un copil in varsta de 3 ani, cu bronhopneumonie si insuficienta respiratorie cronica acutizata (dispnee mixta cu debut din perioada neonatala,accentata la efort si in timpul intercurentelor respiratorii). Pe baza datelor anamnestice, clinice si paraclinice, corelate cu evolutia trenanta a simptomatologiei respiratorii, se decide efectuarea bronhoscopiei ce releva compresie extrinseca bronhie principala stanga si ecografiei cardiace la care se remarca compresie de cavitati cardiace drepte, motiv pentru care se impune efectuarea angio-CT ce evidentiaza hernie hepatica anterioara si stenoza de bronhie principala stanga distal secundara pozitionarii intre aorta descendenta si ramul stang al arterei pulmonare. In urma consulturilor interdisciplinare se stabileste oportunitatea interventiei chirurgicale, ulterior cu evolutie favorabila.

RARE CASE OF RESPIRATORY DISTRESS IN A CHILD WITH DIAPHRAGMATIC LIVER HERNIA

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Congenital diaphragmatic hernia (HDC) is one of the most common malformations in newborns and occurs in 1 in 2500-5000 births. Boys are more often affected than girls by a ratio of 3: 2. The most common type of HDC is represented by a posterolateral defect (foramen Bochdalek) and is responsible for about 85% of cases.

HDC right (foramen Morgagni) is less common (3%) and is asymptomatic in children, being responsible for the delay in diagnosis. Liver herniation through the foramen Morgagni can exert compression on ipsilaerale intrathoracic organs. Presentation spectrum ranges from acute respiratory failure at birth, to minimal or absent symptoms that may occur much later in life, and represents a diagnostic challenge.

We report a case of diaphragmatic liver hernia, found in a child, age 3 years old, with bronchopneumonia and exacerbations of chronic respiratory failure (dyspnea mixed onset of neonatal period, emphasis during effort and intercurrent respiratory illnesses). On the basis of historical data, clinical and laboratory findings, correlated with persistent respiratory symptoms, we decided to perform bronchoscopy witch revealed main left bronchus extrinsic compression, cardiac ultrasound revealing right heart cavities compression, that led to performing angio-CT witch showed liver herniation and stenosis of the left main bronchus distal, secondary positioned beteen the descending aorta and a branch of the left pulmonary artery. Following interdisciplinary checkups the opportunity for surgery is established, subsequently with favorable evolution.

43. PROVOCARE DE DIAGNOSTIC IN HIPONATREMIA SUGARULUI

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Prezentam cazurile a 2 frati, de 6 luni si 1an si 9 luni, cu internari repetate din perioada de nou nascut/ sugar, pentru sindrom de deshidratare acuta si acidoza metabolica, hiponatremie, hiperpotasemie, cu evolutie clinica dificila, trenanta. Ambii prezinta hipostatura, varsta osoasa intarziata, fara ambiguitate sexuala, tatal cu hipostatura.

S-au exclus posibile cauze ale unui sindrom de pierdere de sare

- insuficienta antehipofizara
- insuficienta corticosuprarenala
- deficite enzimatice
- hiperplazia suprarenala congenitala
- nefropatie cu pierdere de sare
- malformatii renale
- infectii

si ale hipostaturii:

- deficit de hormon de crestere,
- hipotiroidie

Initial s-a suspicionat sindrom adrenogenital, tratat cu glucocorticoizi si mineralocorticoizi. Lipsa de raspuns la tratament a determinat investigatii extensive.

Valorile crescute ale aldosteronului plasmatic, acidoza metabolica, hiponatremia, sodiul urinar normal/ crescut, contextul familial si cresterea staturala deficitara, au dus la stabilirea diagnosticului final de pseudohipoaldosteronism tip 1-forma renala. Pseudohipoaldosteronismul este o boala rara (1:166000), caracterizata prin rezistenta la actiunea aldosteronului, la care se disting 2 forme: tipul 1 -renal AD si stemic AR, tipul 2 cu debut tardiv. Afectiunea este tranzitorie, simptomele dispar dupa varsta de 2 ani, pe masura ce aparatul tubular renal se matureaza, desi aldosteronul plasmatic ramane ridicat.

Raritatea bolii reprezinta o provocare pentru clinician , lipsa de raspuns la terapia cortizonica prelungita fiind o capcana de diagnostic.

Evolutia cazurilor a fost favorabila. Unul din pacienti este asimptomatic cu dieta normala de la varsta de 1 an, iar celalalt raspunde la suplimetarea sodiului in dieta.

RARE CAUSE OF INFANT HYPONATREMIA

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We report the ctwo cases of siblings, aged 6 moths and 1year 9 months, with multiple hospitalizations during the newborn/ small infant period, for acute dehydration syndrome with metabolic acidosis, hyponatremia, hyperkalemia, with difficult clinical course. Both children associate short stature, delayed bone age, no sexual ambiguity, father with short stature.

We excluded other causes of salt deficiency syndrome:

- pituitary insufficiency
- adrenal insufficiency
- enzyme deficiency
- congenital adrenal hyperplasia
- salt deficiency nephropathy
- renal malformations
- infections

and of short stature

- growth hormone deficiency
- hypothyroidism

There was a suspicion for adrenogenital syndrome, and the patients were treated with glucocorticoids and mineralocorticoids. The lack of response determined further investigation.Based on high plasmatic aldosterone levels despite treatment, metabolic acidosis, hyponatremia, normal/elevated urinary sodium, family context and short stature, the diagnosis of pseudohypoaldosteronism type 1- renal was made.

Type 1 pseudohypoaldosteronism is a rare disorder (1:166000), characterized by resistance to aldosterone action, classified into renal type 1 AD and systemic type 1 AR, and type 2 (adolescent hyperkalemic syndrome)

The disease tends to be transient, symptoms resolve in patients older than 2 years. A progressive decrease in urinary salt wastage occurs as the renal tube matures throughout infancy. Older children may be asymptomatic with normal salt intake, but plasma aldosterone remains elevated.

Persistent hyponatremia nonresponsive to systemic corticosteroidsis a challenge. This should determine further investigations, especially when confronting with a rare disease.

Our patients responded well to salt supplementation, one became asymptomatic with normal diet.

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44. FACTORI CE INFLUENȚEAZă CREȘTEREA FRECVENȚEI OTITEI MEDII ACUTE

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Introducere: Otita medie acută (OMA) reprezintă afecțiunea cu cel mai mare grad de adresabilitate la medic în perioada micii copilării. În ultimii ani clinica noastra s-a confruntat cu o creștere a numărului de cazuri de OMA. Obiectiv: Analiza factorilor ce pot influența creșterea frecvenței OMA. Material și metodă: Am studiat retrospectiv fișele pacienților cu vârste cuprinse între 1 și 24 luni, internați în Clinica II Pediatrie, Timișoara, cu diagnosticul de OMA în perioada 1 ianuarie 2013- 31 decembrie 2014. Am obținut un lot de 59 de pacienți pentru care am analizat următorii parametrii: sex, rang, vârsta gestațională, modalitatea nașterii (vaginală versus operație cezariană), greutate la naștere și actuală, statusul vaccinărilor, prescrierea recentă de antibiotic. Rezultate: Majoritatea copiiilor diagnosticați cu OMA au fost născuți prin operație cezariană (33.9% vs 66.1%, p= 0.018). Cei mai mulți dintre copiii cu OMA au urmat tratament antibiotic în istoricul recent (27.12% vs 72.88%, p= 0.001). 83.5% din pacienți au fost vaccinați conform schemei Ministerului Sănătății, niciunul dintre copii nefiind vaccinat antipneumococic. Concluzii: În lotul nostru, frecvența crescută a operației cezariene și a tratamentului antibiotic recent precum și lipsa vaccinării antipneumococice par a fi responsabile de creșterea numărului de OMA. Sunt necesare studii de anvergură pentru a confirma aceste ipoteze.

FACTORS INFLUENCING THE INCREASED FREQUENCY OF ACUTE OTITIS MEDIA

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Background: Acute otitis media (AOM) is the condition with the highest degree of medical addressability during early childhood. In recent years our department has faced a growing number of cases with AOM. Objective: Analysis of the factors that can influence the growing numbers of children presenting with AOM. Methods: We studied retrospectively records of patients aged 1 to 24 months, admitted with AOM to the 2nd Pediatrics Clinic, between January 1st 2013 and December 31st 2014. Our group consisted of 59 patients for whom we analyzed the following parameters: gender, birth rank, gestational age, type of birth (vaginal versus caesarean section), birth weight, current vaccination status, and recent antibiotic use. Results: Most children diagnosed with AOM were born through caesarean section (33.9% vs 66.1%, p = 0.018). For most children with AOM antibiotics were used in recent history (27.12% vs 72.88%, p = 0.001). 83.5% of patients were immunized according to the Ministry of Health schedule, while none of the children were vaccinated against Streptococcus Pneumoniae. Conclusions: In our study, increased frequency of caesarean section, recent antibiotic therapy and the lack of

pneumococcal vaccination seem to be responsible for the increasing number of AOM. Large-scale studies are needed to confirm these hypotheses.

45. FALLOT UP RMN VERSUS ECHOCARDIAC

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Tetralogia Fallot- malformatie de cord cianogena, este cunoscuta in literatura de specialitate ca prototipul malformatiilor cardiace cianogena complexe. In prezent s-au dezvoltat tehnici imagistice performante
(parametrii echografici, CT si RMN cardiac), care permit stabilirea diagnosticului, prognosticului, timpului operator optim si urmarirea pe termen lung a rezultatului postoperator.
Va prezentam cazul unei adolescente de 17 ani, diagnosticata la nastere cu Tetralogie Fallot forma severa (cu dextropozitia aortei aproape 100%, incadrabila in tipul ventricul drept cu cale dubla de iesire). A fost operata per primam la varsta de 2 ani. Dupa 15 ani posteoperator prezinta regurgitare tricuspidiana si pulmonara semnificativa, deteriorare marcata a functieiei ventriculare drepte, in discordanta cu starea clinica foarte buna.
Caracterul bizar echocardiographic, malaliniamentul aorta- ventricul stang, morfologia complexa au facut necesar evaluarea prin RMN cardiac. Coroborand datele clinice, anamnestice, imagistice s-a stabilit prognosticul, conduit terapeutica si chirurgicala ulterioara.
Concluzii: RMN cardiac este gold standardul imagistic pentru evaluarea ventriculului drept. Prognosticul pre/postoperatot in Tetralogia Fallot este apreciat prin markeri echocardiografici moderni si prin RMN cardiac. Postevaluare RMN, cazul prezentat are indicatie de reinterventie chirurgicala cu reconstructia tractului de ejectie al ventriculului drept.
Cuvinte cheie: Tetralogie Fallot, Echocardiac, RMN cardiac

FALLOT UP MRI VERSUS ECHOCARDIOGRAPHY

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Tetralogy of Fallot- cyanotic heart malformation - is known in the literature as the prototype of complex cyanotic heart malformations. Current developments in advanced imaging techniques (ultrasound parameters, cardiac CT and MRI), allow for diagnosis, prognosis, and optimal surgery time calculation as well as tracking long-term outcome after surgery.

We present the case of a 17 years old teenager, diagnosed with the severe form of Tetralogy of Fallot at birth (with right-sided aortic arch almost 100%, classed as right ventricular type with double track output). She underwent her first surgery at age 2. 15 years after the surgery the patient has significant tricuspid and pulmonary regurgitation and also marked deterioration of the right ventricular function witch contrast to the good clinical condition.

The bizarre echocardiographic character witch revealed the strange alignment between the aorta and left ventricle as well as the complex morphology, made necessary cardiac MRI assessment. Corroborating clinical data, anamnesis, and imaging was established prognosis, therapeutic and surgical subsequent conduct.
Conclusions: Cardiac MRI imaging is the gold standard for evaluating the right ventricle. The before/after surgery prognosis in the Tetralogy of Fallot is established by using modern echocardiographic markers and cardiac MRI. After the MRI revue the case has a strong indication for subsequent surgery for the reconstruction of the right ventricle outflow path.
Key wors: Tetralogy Fallot, echocardiogaphy, cardiac MRI

46. DIAGNOSTICUL ŞI MANAGEMENTUL, PARAMETRII CU IMPACT MAJOR ASUPRA PROGNOSTICULUI CANALULUI ATRIOVENTRICULAR COMUN

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Introducere: Canalul atrioventricular comun (CAVC) este o malformație congenitală cardiacă, ce poate varia de la simplu la complex, cu management și evoluție individualizate.
Scopul lucrării este reprezentat de evaluarea diagnosticului și managementului copiilor cu CAVC; parametrii cu impact major asupra prognosticului.

Materiale şi metode: Am realizat un studiu retrospectiv pe un grup de 31de pacienți cu CAVC, monitorizați ecocardiografic în Clinica II Pediatrie Timișoara, în perioada 2010-2015.
Rezultate: Ambele sexe sunt afectate în egală măsură, 38,7% din pacienți au sindromul Down, cifră comparabilă cu literatura de specialitate. Au predominat cazurile diagnosticate sub 1an, fapt corelat cu urmărirea sarcinii, echipament adecvat și personal medical bine pregatit.
Majoritatea pacientilor au beneficiat de corectie totală primară, o parte a necesitat intervenție paleativă cu corecție ulterioară, iar un numar mic doar de banding de arteră pulmonară. La cei cu forme parțiale sau tranziționale supraviețuirea este de 100%, dar scade la cei cu formă completă, cu cât crește gradul de complexitate a leziunilor, deoarece formele severe au restanțe postoperator, unele necesitând reintervenție.
Concluzii: Idealul diagnostic este reprezentat de diagnosticul prenatal. Întarzierea diagnosticului duce la întarzierea tratamentului chirurgical cu alterarea prognosticului prin apariția complicațiilor. Evolutia este influențată de tipul defectului, formele complexe fiind tehnic dificil de corectat, cu prognostic mai puțin favorabil.
Cuvinte cheie: diagnostic, prognostic, malformatie cardiaca congenitala

DIAGNOSIS AND MANAGEMENT, MAJOR IMPACT PARAMETERS IN ATRIOVENTRICULAR SEPTAL DEFECT

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Introduction: Atrioventricular septal defect(AVSD) is a congenital heart defect, ranging from simple to complex, each form with different management and evolution. Our aim is to assess the diagnosis and management of children with AVSD as parameters with major impact on the prognosis.
Materials and Methods:We contucted a retrospective study including 31pacients of Clinic II Pediatrics Timisoara having AVSD, during follow-up echocardiographic examinations, between 2010-2015.
Results:Both sexes were equally affected and 38,7% of all pacients had Down syndrome, number comparable to literature.Most cases were diagnosed under age1, correlated with pregnancy follow-up, adequated equipment and well trained personnel.The majority of pacients underwent total primary correction,some needed palliative surgery with subsequent correction and a small number benefited only of pulmonary artery banding. Some are yet to undergo surgery.Survival is 100% in the partial or transitional form,but it decreases in the complete form in direct proportion with the complexity of lesions, because severe impared forms lead to surgical backlogs,some requiring reintervention.
Conclusions: The ideal diagnosis is the prenatal diagnosis. Delay in diagnosis leads to delaying surgical treatament,thus altering the outcome due to complications. The evolution depends on the type of defect:complex forms,are technically more difficult to correct and therefore with less favorable prognosis.
Key words: diagnosis, prognosis, congenital heart defect

POSTERE / POSTERS

ALERGOLOGIE / ALERGOLOGY

1. PROFILAXIA INFECTIILOR VIRALE LA COPII CU BOLI ALERGICE

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USMF Nicolae Testemitanu, Chisinău, Republica Moldova

Introducere. Infecţiile virale acute (IV) la copii prezintă o problemă majoră pentru sănătatea publică. Scopul lucrării a fost aprecierea eficacitatii preparatului Kagocel ® (NEARMEDIC PLUS, Rusia) in tratamentul şi profilaxia infecţiilor virale (IV) la copii cu maladii alergice. În studiu au fost incluşi 123 copii cu vârstele de la 3 pina la 15 ani, dintre care: 50- cu astm bronşic (AB), 83- cu dermatită atopică (DA), 60 – cu rinita alergică (RA). La terapia de bază in perioada de toamna-iarna copii au primit o cură de tratament profilactic cu KAGOCEL ® (cicluri de 7 zile). De menţionat că la toţi pacienţii s-a micşorat frecvenţa IV până la 1-2 per sezon, la 40 copii cu astm bronşic (80%) s-a redus numărul de pusee de la 1-2 pe săptămâna până la 1-2 în trimestru. La 70 de copii cu DA (84%) nu s-a acutizat maladia iar la 12 copii (15 %) – acutizarea a fost locală şi de scurtă durată. La 51 de copii cu RA (85%) s-a notat remisia clinică.
Concluzie: Datele obţinute au demonstrat efectul favorabil profilactic al medicamentului Kagocel in frecventa şi severitatea IV la copii cu maladii alergice.

THE PROPHYLAXIS OF VIRAL INFECTIONS IN CHILDREN WITH ALLERGIC DISEASES

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Introduction. The acute viral infections (VI) present a major problem for public health. The goal of work was the treatment and prophylaxis of viral infections (VI) in children with allergic diseases through the using of Kagocel ® (NEARMEDIC PLUS, Russia). In the study were included 123 children having the age from 3 till 15 years, 50 of which – with bronchial asthma (BA), 83 – with atopic dermatitis (AD), 60 – with allergic rhinitis (AR). In autumn-winter period these children have received a cure of prophylactic treatment with Kagocel ® (cycles of 7 days). It should be mentioned the frequency of VI was reduced until 1-2 times per season in all patients. Moreover in 40 patients with BA (80%) the number of exacerbations was reduced from 1-2 per week until 1-2 per trimester. In 70 children with AD (84%) there were not exacerbations of main disease and in 12 (15%) exacerbation was local and has short duration. In 51 children with AR (85%) the clinical remission has been noted.
Conclusion: The obtained data have demonstrated the favorable prophylactic effect of Kagocel on the frequency and severity of VI in children with allergic diseases.

2. TOXOCAROZA FORMA SEVERA LA COPIL - DIFICULTATI DE DIAGNOSTIC

Singer Cristina[1], Petrescu Ileana[1], Cosoveanu Simona[1], Oancea Gabriela[2], Dinca Andreia[2], Dumitru Bianca[2], Cristea Camelia[2], Dragomir Flavius[2]
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Este prezentat cazul unui copil in varsta de 1 an si 6 luni, sex feminin, G= 9Kg, din mediul rural, internat in clinica noastra in februarie 2015 pentru febra, tuse. Datele anamnestice, examenul clinic si radiologic au condus initial la diagnosticul de pneumonie acuta interstitiala. Pe parcursul internarii a prezentat repetate exacerbari cu manifestari de insuficienta respiratorie acuta, in afebrilitate. Examenele paraclinice au relevant anemie (Hb=8,4g%), leucocitoza (L=13300/mm3, NS=57%, Ly=37%, M=6%), teste inflamatorii cu valori mult crescute, sideremie, calcemie scazute, teste hepatice, renale, examen coproparazitar, ECHO abdominal normale, electroforeza proteinelor serice – hiperproteinemie (11,4g%) cu hipergamaglobulinemie (31,7%), IgE, IgG cu valori mult crescute.
Am continuat investigatiile pentru precizarea diagnosticului: testul sudorii, CIC, AAN, FR, C3, testul TORCH, ECHO cord normale, atg HBs, atc antiHVC, test ELISA pentru HIV negative, examen oftalmologic relatii normale. Radiografii pulmonare repetate – opacitati interstitiale peribronhovasculare peri si infrahilar drept. Punctia medulara– procent crescut de eozinofile in mai multe stadii de maturatie, in procent de aproximativ 20%, celelalte serii medulare normale, fara celule atipice; hemograma din pulpa degetului a aratat anemie (Hb=7,9g%), leucocitoza (L=14000/mm3, NS=17%, E=36%, Ly=42%, M=5%). Se reorienteaza diagnosticul spre o posibila Toxocaroza - atc anti toxocara canis=55,6 NTU. In a 3-a saptamana de internare pacienta a prezentat scurte crize de convulsii afebrile, CT cranian, EEG normale.
Copilul s-a externat dupa 5 saptamani de spitalizare cu diagnosticul de Toxocaroza forma severa cu manifestari pulmonare si cerebrale, cu tratament antiparazitar, si a ramas in evidenta clinicii noastre.

SEVERE TOXOCARIASIS IN CHILDREN – DIAGNOSTIC DIFFICULTIES

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We present the case of an 18-month-old girl, weighing 9 kg, from rural area, admitted to our clinic in February 2015 for fever and cough. The anamnestic data and the clinical and radiological examinations initially suggested the diagnosis of acute interstitial pneumonia. During hospitalization, she repeatedly presented exacerbations with acute respiratory failure, but without fever. Paraclinic examinations revealed anemia (Hb=8.4g%), leukocytosis (L=13300/mm3, NS=57%, Ly=37%, M=6%), inflammatory tests with highly increased values, low values of serum iron and serum calcium, normal hepatic and renal tests, coproparasitary examination, and abdominal ultrasound, serum protein electrophoresis - hyperproteinemia (11.4g%) with hypergammaglobulinemia (31.7%), the values of IgE and IgG being highly increased.

We continued our investigations to establish a diagnosis: the sweat test, ICC, ANA, RF, C3, TORCH test, and the heart ultrasound were normal, atg HBs, atc antiHVC, ELISA test for HIV were negative, normal ophthalmologic examination. Repeated pulmonary x-rays – peribronchovascular interstitial thickening, at the level of peri- and right infrahilar area. Medullary puncture – increased percentage of eosinophils in several stages of maturity, approximately 20%, normal values for the other medullary series, without atypical cells; the fingertip hemogram showed anemia (Hb=7.9g%), leukocytosis (L=14000/mm3, NS=17%, E=36%, Ly=42%, M=5%). The diagnosis turned to a possible toxocariasis - atc anti toxocara canis=55.6 NTU. In the third week of hospitalization, the patient presented short crises of nonfebrile seizures. Normal CT and EEG brain.

The child was discharged after 5 weeks, the diagnosis being severe toxocariasis with pulmonary and neurological manifestations; she was recommended an antiparasitary treatment and remained in our clinic's records.

3. THE QUALITY OF LIFE - AN INDICATOR OF FAIR TREATMENT OF ALLERGIC RHINITIS IN ADOLESCENTS

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Aims: The purpose of this study was to estimate the quality of life of adolescents with allergic rhinitis and to establish the best treatment.

Material and method: The study was performed on 42 adolescents (aged between 12 and 18 years) admitted to a university children's hospital between 1 January 2013 and 31st December 2014. To assess quality of life we used five symptoms score and visual analog scale. Statistical processing was performed by Student's t-test.

Results: Regarding the sex distribution of patients 42% of the cases were girls and 58% were boys. Depending on severity score, patients were divided into two groups: 26% of patients with mild persistent allergic rhinitis and 74% of cases with moderate-severe persistent allergic rhinitis. After the first week of treatment, 80% of the 31 patients with moderate-severe persistent allergic rhinitis recognized a net improvement of symptoms, with a good quality of life without affecting daily activities and sleep. 10% of the 31 patients with moderate-severe persistent allergic rhinitis continued to maintain the source of allergens (cats and dogs) in the living environment. 7% of patients have not regularly administered treatment, being without family support.

Conclusions: Patients' quality of life depends on the time of diagnosis, the promptitude of establishing treatment and allergen avoidance. Moderate-severe persistent allergic rhinitis significantly affects the quality of life. The quality of life is more affected as well as the total score of symptoms is higher.

Key words: allergic rhinitis, adolescent, life

BOLI BOLI INFECTIOASE ȘI VACCINOLOGIE/ INFECTIOUS DISEASES AND VACCINOLOGY

4. TOXOCAROZA IN PATOLOGIA PEDIATRICA. CAZURI CLINICE

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Toxocaroza este o zoonoză cauzată de ingestia de larvele *Toxocara canis* /*Toxocara cati*. Scop. Autorii mentioneaza particularitatile diagnostice la doi copii transferati in contextul suspiciunii de leucemie acuta. Material si metode. Primul caz: baiat in varsta de 2 ani internat pentru astenie marcata, febra, status nutritional deficitar, paloare tegumentara, hepato-splenomegalie. Cazul 2: baiat de 3 ani cu paloare tegumentara, retard de crestere. Rezultate. Investigatii de laborator: leucocitoza (53.000/mm^3 respectiv 104.000/mm^3) cu eozinofilie si anemie (ambele cazuri); transaminase crescute cu serologie pozitiva pentru virusul Epstein-Barr (primul caz); imunofenotiparea sangelui periferic a aratat predominanta limfocitelor T CD4+ (cazul 2); examenul maduvei osoase a identificat hiperplazia precursorilor eozinofilici (ambele cazuri) justificand suplimentarea investigatiilor ce au relevat titru crescut de anticorpi anti-*Toxocara canis* (toxocaroza confirmata) si serologie negativa pentru *Toxoplasma gondii*/ *Taenia Echinococcus*. Examenul coproparazitologic: negativ (cazul 1) si pozitiv la al 2-lea la care s-a decelat coinfestatia parazitara (oua *Trichocephalus trichiuris* /*Ascaris lumbricoides*, chisturi *Giardia intestinalis)*. Evaluarea imagistica nu a aratat modificari patologice. Examenul oftalmologic a fost in limite normale (ambele cazuri). Diagnosticul diferential a inclus leucemia acuta eozinofilica, dar examenul maduvei osoase a exclus boala. Pacientii au fost tratati cu derivati benzimidazolici cu rezolutia prompta a leucocitozei. Concluzii. 1.Autorii au prezentat 2 cazuri cu toxocaroza asociata cu anomalii hematologice si infectii /infestatii: mononucleoza infectioasa (primul caz) si co-infestatie parazitara pentru al 2-lea caz (ascaridiaza, giardiaza, tricocefaloza). 2.Leucocitoza cu eozinofilie poate reprezenta un marker pentru toxocaroza. 3.Pe fondul leucocitozei, autorii au considerat leucemia acuta, justificandu-se analiza maduvei osoase.

TOXOCARIASIS IN CHILDREN. CLINICAL CASES

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Background and aims. Toxocariasis is helminth infection of humans caused by the roundworms *Toxocara canis* and *Toxocara cati*. The authors emphasize diagnosis particularities in 2 children transferred in pediatric department for further evaluation in context of acute leukemia suspicion. Methods. First case: 2 year-old boy with fatigue, fever, impaired nutritional status, skin pallor, hepatosplenomegaly. Second case: 3 year-old boy with skin pallor and failure to thrive. Results. Laboratory investigations shown leukocytosis (>50,000/mm^3), eosinophilia and anemia (both cases); liver function impairment and positive serology for Epstein-Barr virus (first case); peripheral blood flowcytometry shown predominance of T lymphocytes CD4+ (second case); bone marrow exam identified eosinophil precursors hyperplasia (both cases) justifying additional tests that revealed high serum titers for *Toxocara canis* and negative serology for *Toxoplasma gondii*/ *Echinococcus*. Stool exam: negative (first case) and parasitic co-infection (second case) revealing *Trichocephalus trichiuris* eggs, *Ascaris lumbricoides* eggs and *Giardia intestinalis* cysts. Imagistic evaluation (abdominal ultrasonography, echocardiography) was normal. No ophthalmological anomalies in both children. Main differential diagnosis was focused on acute eosinophilic leukemia (in context of leukocytosis, eosinophilia) and bone marrow examination excluded it. The patients received benzimidazole compounds with prompt leukocytosis improvement. Conclusions. 1.The authors have presented two cases with hematological peculiarities and concomitant infections: toxocariasis associated with infectious mononucleosis (first case) and parasitic co-infections in second case (toxocariasis, ascaridiasis, giardiasis, trichuriasis); 2.Eosinophilic leukocytosis could represent a reliable marker for *Toxocara* infection in children; 3.Because of leukocytosis authors considered acute leukemia in both cases, justifying bone marrow analysis.

5. OSTEOMIELITA ACUTA SI ARTRITA SEPTICA – ETIOLOGIE SI MANAGEMENT

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Introducere: infectiile osteo-articulare reprezinta o cauza majora de morbiditate la copii.
Obiectiv: evaluarea etiologiei, managementul si rezistenta la antibiotice in osteomielita acuta si artrita septica.
Metode: studiu retrospectiv pe un lot de copii cu varste intre o luna si 16 ani internati in Spitalul Clinic de Copii Brasov in intervalul 2005-2014.
Rezultate: varsta medie a fetelor afectate de osteomielita acuta este de 7.4 ani, iar a baietilor de 10.3 ani versus artrita septica, unde varsta medie a baietilor este de 5.7 ani, iar cea a fetelor 6.89 ani. 55% dintre copiii cu osteomielita acuta au provenit din mediul rural. Procentele pentru mediul de provenienta al pacientilor diagnosticati cu artrita septica au fost impartite in mod

egal. 75% dintre baieti au fost diagnosticati cu osteomielita acuta si 64.2 % cu artrita septica. Dintre pacientii diagnosticati cu osteomielita acuta, carora li s-au efectuat culturi din lichidul articular, majoritatea s-au pozitivat cu *Staphylococcus aureus.* In cazul artritei septice, culturile din lichidul articular au fost in marea lor majoritate negative, dintre cele pozitive principalul agent etiologic a fost *Staphylococcus aureus*, multirezistent la antibioterapie (Penicilina, Ampicilina, Augmentin, Piperacilina-tazobactam, Oxacilina, Ceftazidime, Cefotaxime, Cefuroxime, Ceftriaxon, Eritromicina, Claritromicina, Teicoplanin). Rezistenta la antibioterapie a facut ca media de spitalizare pentru osteomielita acuta sa depaseasca 22 de zile, iar in cazul artritei septice 14 zile.
Concluzii: *Staphylococcus aureus* ramane cel mai comun patogen implicat in aparitia artritei septice si osteomielitei acute.

ACUTE OSTEOMYELITIS AND SEPTIC ARTHRITIS – ETIOLOGY AND MANAGEMENT

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Background: osteoarticular infections represent a major cause of morbidity in children.
Aim of study: assessement of etiology, management and antimicrobial resistance in acute osteomyelitis and septic arthritis at children in Brasov county.
Methods: we conducted a retrospective study at children aged between one month and 16 years admitted to the University Childrens Hospital Brasov, Romania between 2005 and 2014.
Results: the average age of girls with acute osteomyelitis is 7.4 years and of the boys is 10.3 years versus septic arthritis, in which age average of boys is 5.7 years and that of the girls is 6.89 years. 55% of the children with acute osteomyelitis came from rural environment. Percentages regarding the living environments of patients diagnosed with septic arthritis were equally distributed. 75% of the boys were diagnosed with acute osteomyelitis and 64.2% with septic arthritis. Among the patients with acute osteomyelitis who underwent synovial fluid examination, the majority of the cultures were positive for *Staphylococcus aureus.* In the case of septic arthritis, the majority of synovial fluid cultures were negative, the positive ones were with *Staphylococcus aureus* resistant to Penicillin, Ampicillin, Amoxycillin-Clavulanate, Piperacillin-Tazobactam, Oxacillin, Ceftazidime, Cefotaxime, Cefuroxime, Ceftriaxone, Erythromycin, Clarithromycin, Teicoplanin. Antimicrobial resistance was responsible for a length of hospitalisation above 22 days for osteomyelitis and 14 days for septic arthritis.
Conclusions**:** *Staphylococcus aureus* remains the most common pathogen of septic arthritis and acute osteomyelitis.

6. GASTROENTEROCOLITELE ACUTE NEBACTERIENE - CONSIDERAȚII CLINICO-ETIOLOGICE PE UN LOT DE 60 DE COPII

SL Dr. Dumitra Simona, Asist. Univ. Dr. Carmen Crișan, SL. Dr. Luminița Pilat, stud. Daniela Ciupea
Universitatea de Vest "Vasile Goldiș" Arad

Scop – de a identifica germenii determinanți și principalele caracteristici clinice ale gastroenterocolitelor nebacteriene.
Material și metodă- Lotul a constat din 60 de copii internați în perioada 01.01.2015-31.03.2015 cu diagnosticul de gastroenterocolită acută și coproculturi negative. Au fost elaborate chestionare pentru părinți referitoare la simptomatologie, precum și o fișă medicală personalizată completată de medicii din studiu.Toți copii au fost screenați pentru Rotavirus, Norovirus și paraziți (metoda Elisa rectiv Ridascreen generația III și examen coproparazitologic).
Rezultate-Au predominat categoriile de vîrsta 0-1 an-46,66% și 1-3 ani-36,66%, față de 3-6 ani-10% , 6-16 ani-6,66% pacienți. Rotavirusul a fost identificat în 20% cazuri, Norovirusul în 15%, Giardia Lamblia 11,66%, iar în 51,66% cazuri nu a fost stabilită etiologia. Tabloul clinic a fost dominat de vărsături 78,33%, scaune apoase 85%, dureri abdominale 38,33%, febră 26,66%. S-au menționat în procente mici asocieri cu scaune cu striuri sangvinolente 8,33%, IACRS 8,33%, rash 5%, convulsii 1,66%.
Concluzii
La peste 50% din copiii lotului nu a fost identificată etiologia diareei.
Introducerea de rutină a unor teste rapide pentru Rotavirus, Norovirus, Adenovirus este oportună pentru evitarea introducerii de antibiotice într-o diaree nebacteriană.
Modelul vărsături frecvente urmate de diaree apoasă este sugestivă pentru o etiologie nebacteriană, justificând excluderea obligativității coproculturilor, cu scăderea costului spitalizării.
Lucrare efectuată în cadrul grantului intern PI/4 al Universității de Vest "Vasile Goldiș" din Arad
Cuvinte cheie - gastroenterocolite, copil, etiologie, tablou clinic.

NONBACTERIAL ACUTE GASTROENTEROCOLITIS – CLINICAL –ETIOLOGICAL CONSIDERATIONS ON 60 CASES

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Purpose – to identify the main germs and clinical traits of nonbacterial gastroenterocolitis.

Material and method – The lot consisted of 60 children hospitalized in the Department of Pediatrics of Arad between 01.10.2014 – 31.03.2015 with negative coprocultures. Questionnaires for the parents regarding the symptoms were elaborated, as well as a personalized medical record completed by physicians. Children were screened for Rotavirus, Norovirus and parasites (Elisa method with reactive Ridascreen third generation and parasitological examination)
Results – The predominant age categories where those of 0-1 years- 46,66% and 1-3 years 36,66%, beside 3-6 years-10%, 6-16 years – 6,66% pacients. The Rotavirus was identified in 20% of the cases, the Norovirus in 15 %, Giardia Lamblia in 11,66 %, an in 51,66% of the cases the ethiology was not established. The clinical picture was dominated by vomiting 78,33%, watery stools 85%, abdominal pain 38,33%, fever 26,66%. . There where mentioned in smaller percentages associations with bloody stools 8,33%, runny nose 8,33%, rash 5%, seizures 1,66%.
Conclusions
At more than 50% of the children, the etiology of the diarrhea was not identified.
Routine introduction of some rapid tests for Rotavirus, Norovirus is oportune in order to avoid the use of antibiotics in a nonbacterial diarrhea.
The model of frequent vomiting followed by watery stools is suggestive for a nonbacterial etiology .
Work carried out within the internal grant PI/4 of the "Vasile Goldiş" Western University of Arad, Romania
Key words- gastrenterocolitis, child, etiology, clinical aspects

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7. ADENOMEGALIE AXILARA - TOXOCARA CANIS VERSUS EPSTEIN-BARR

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Introducere
Toxocara este o zoonoza parazitara, data de infestatia omului cu larve de Toxocara canis. Debutul bolii depinde de intensitatea invaziei, raspandirea larvelor in organe si tesuturi, frecventa reinfestarilor si de statutul imun al gazdei. Formele clinice de toxocaroza includ forma generalizata majora si minora, forma localizata oculara si neurologica.

Obiective
Prin aceasta lucrare ne dorim aducerea in prim-plan a unei manifestari clinice mai rar intalnite in cadrul unei toxocaroze, o forma generalizata minora.
Material si metoda
Subliniem etapele diagnostice si evolutia clinica a unui pacient în varsta de 5 ani care s-a prezentat in serviciul pediatrie cu o adenomegalie axilara dreapta izolata. Investigatiile serodiagnostice deceleaza anticorpi IgG Toxocara canis 340 unitati NovaTec, in conditiile in care valoarea de referinta, pentru un rezultat negativ, era mai mica de 9 si niveluri crescute de anticorpi VCA IgG Epstein Barr. Raspunsul major al gazdei la antigenele parazitului include o marcata eozinofilie si o hipergammaglobulinemie IgE.
Concluzii
Adenomegalia benigna axilara s-a datorat unei limfoprolifereri reactive.
Biopsia chirurgicala ar fi fost obligatorie cand etiologia nu era stabilita prin investigatiile obisnuite sau evolutia clinica sub tratament ar fi fost nefavorabila.
In tara noastra, nu exista date statistice, infestarea cu toxocara canis fiind raportata sporadic. Boala la om poate fi prevenita, prin deprinderea unor reguli de igiena; se impune cunoasterea si depistarea zoonozei in vederea tratamentului corect.
Cuvinte cheie: Toxocara canis, adenopatie, Epstein Barr

AXILLARY ADENOMEGALLY – TOXOCARA CANIS VERSUS EPSTEIN-BARR

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INTRODUCTION
Toxocara is a parasitic zoonosis, human infestatia date with larvae of Toxocara canis. The onset of the disease depends on the intensity of the invasion, evolution larvae in bodies and tissue, the reinfestation frequency and the immune status of the host. Clinical toxocaroze provides the generalized form of this disease major and minor, localized ocular and neurological.
OBJECTIVES
In this presentation we wish to bring to the foreground one of the clinical manifestations of a rarely encountered in a toxocaroze, a minor generalized form.
MATERIAL AND METHOD

We point out the stages of development and clinical diagnoses of a 5 years old patient who presented in the pediatric service with an isolated right axillary adenomegally. The serodiagnostic investigations can detect the antibodies IgG Toxocara canis 340 units NovaTec, in the conditions in which the reference value for a negative result was less than 9 and increased levels of Epstein Barr VCA IgG antibodies. The major answer of the host's parasite antigens includes an eosinophilia and IgE hipergammaglobulinemie.

CONCLUSIONS

The adenomegally benign axillary was due to a reactive limfoproliferation. The surgical biopsy would be required when the etiology wasn't fixed through the standard investigations or clinical evolution under treatment would have been unfavorable. In our country, there aren't any statistic dates, the toxocara canis infestation being sporadically. The disease at humans can be prevented through acquiring strict hygiene rules.

KEY WORDS: ADENOMEGALLY, TOXOCARA CANIS, EPSTEIN-BARR

8. SEPSISUL LA COPIL – CONSIDERATII PE BAZA A DOUA CAZURI CLINICE

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Introducere: Sepsisul reprezinta o modalitate de evolutie a unei infectii, cu o rata de mortalitate de aproximativ 10% din cazuri. Prima definitie universala de sepsis, sepsis sever si soc septic a fost stabilita in anul 1991 si permanent revizuita. In 2011 Clutton-Brock defineste sepsisul ca Sindromul de răspuns inflamator sistemic (SIRS) de cauză infecţioasă certă sau probabilă. Pentru a defini Sindromul de răspuns inflamator sistemic este necesara prezenţa a cel puţin două din următoarele criterii: temperatură mai mare de 38C sau mai mică decât 36C, tahicardie, tahipnee, leucocitoza.

Material si metoda: Lucrarea de fata prezinta doua cazuri de septicemie la copii internati in clinica noastra.

Un sugar de 10 luni fara antecedente patologice se interneaza cu febra inalta 39,5-40C, tahicardie, tahipnee. Investigatiile paraclinice au relevat leucocitoza 32.210/mm3 cu neutrofilie, reactanti de faza acuta crescuti, hemocultura pozitiva cu E.Coli. Al doilea copil de 1an si 2 luni se prezinta pentru febra 39-39,5C, tahicardie, sensibilitate otica. Se stabileste diagnosticul de Otita supurata preperforativa, se practica timpanotomie . Dupa 24 de ore starea generala se agraveaza, paraclinic se deceleza leucocitoza cu neutrofilie, reactanti de faza acuta crescuti, hemocultura pozitiva.

Concluzii: Diagnosticul precoce si tratamentul corect instituit reduce complicatiile severe si rata de mortalitate in cazul sepsisului la copil.

Cuvinte cheie: sepsis, copil, otita.

SEPSIS IN CHILDREN - CONSIDERATIONS BASED ON TWO CLINICAL CASES

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Introduction: Sepsis is a way of evolution of an infection with a mortality rate about 10% of cases. The first universal definition of sepsis, severe sepsis and septic shock was established in 1991 and constantly revised. In 2011, Clutton-Brock defines sepsis as a systemic inflammatory response syndrome (SIRS) of definite or probable infectious cause. To define systemic inflammatory response syndrome it is necesary to have the presence of at least two of the following criteria: temperature higher than 38C or lower than 36C, tachycardia, tachypnea, leukocytosis.

Methods: This study presents two cases of sepsis in children admitted to our clinic.

An infant of 10 months old, without medical history was hospitalized with high fever 39.5C - 40C, tachycardia, tachypnea. Laboratory investigations revealed leukocytosis 32,210 / mm3 with neutrophilia, raised acute phase reactants, positive blood culture with E. Coli. The second child of 1 year and 2 months old, was hospitalized with fever 39C - 39,5C, tachycardia, otic sensitivity. The child was diagnosed with otitis media with effusion and a tympanostomy was performed. After 24 hours the general condition worsens, laboratory investigations revealed leukocytosis with neutrophilia, raised acute phase reactants, positive blood culture.

Conclusions: Early diagnosis and proper treatment reduce complications and mortality rate in severe sepsis in children.

Key words: sepsis, children, otitis

9. ASPECTE PARTICULARE IN INFECTIA DE TRACT URINAR LA SUGARI

Dr. Ciurea Rujita Dr. Ciurea Paul Adrian

Definiţie

Infecţia de tract urinar (ITU) se defineşte prin prezenţa unui process inflamator determinat de prezenta microorganismelor la nivelul tractului urinar, caracterizata printr-o bacteriurie ≥105, insotita de manifestari clinice bruste cu hipertermie sau poate fi descoperita intamplator la un examen de rutina al urinei.

Material si metoda.

S-au luat in studiu 40 de sugari(varsta 0-1an), internati in cadrul sectiei Pediatrie 1 in pereioada ianuarie –decembrie 2014.Cazurile au fost selectate pe baza uroculturilor positive, depistate pe parcursul internarii, cu diagnostic clinic la internare altul decat infectia urinara.

Concluzii

1.Particularitatea ITU in perioada de sugar este data de o simptomatologie mascata.

2. Manifestările clinice ale ITU la sugar sunt nespecifice: febră,agitaţie, inapetenţă, vărsături, stagnare în greutate sau spor ponderal negativ, diaree . Aceste simptome pot fi foarte bine atribuite eruptiei dentare ducand la intrazierea diagnosticului.

3. La sugarul cu febră fără o cauză aparentă clinic sau dacă se asociază cu stare toxică şi deshidratare , pe un teren carentat se va suspecta ITU.

4. Cei mai frecventi factori favorizanti : – cei care permit bacteriilor sa invadeze tractul urinar cu un sistem de aparare imatur la aceasta categotie de varsta , au fost fimoza la baieti, igiena deficitara si MPC.

5. Urocultura reprezintă metoda de gold in diagnosticul ITU la sugar.

6. Cele mai frecvente microorganisme implicate in aparitia infectiilor de tract urinar sunt germenii gram negativi: E. Coli, Klebsiella, Enterobacter, Proteus.

7.Antibiograma a relevat o sensibilitate crescuta a germenilor la cefalosporine de generatia a III-a si la polipetide.

8.Este necesara o activitate sustinuta pentru imbunatatirea compliantei la tratament, a deprinderilor de igiena intima, supraveghere diferentiata la domiciliu.

Cuvinte cheie: manifestari atipice-sugar –itu.

PARTICULAR ASPECTS IN URINARY TRACT INFECTIONS

Dr. Ciurea Rujita
Dr Ciurea Rujia, Dr. Ciurea Paul Adrian

Definition

The urinary tract infection is defined as the presence of an inflammatory process, determined by the existence of the microorganisms in the urinary tract, characterized by a bacteriuria ≥105, accompanied by sudden clinical manifestation hyperthermia or can be discovered by chance on a routine examination of urine.

Material and method

They were taken in study 40 infants (age 0-1 year), hospitalized in Pediatrics I department between January and December 2014.Cases were selected by positive urine culture, found during hospitalization, with clinical diagnosis on admission other than urinary infection.

Conclusions

1. The particularity of the urinary tube infection in infancy is given by masked symptoms.
2. Clinical manifestations of UTI in infants are nonspecific: hyperthermia, agitation, anorexia, vomiting, stagnation in weight or negative weight gain, diarrhea. These symptoms can be attributed to dental eruption, resulting in delayed diagnosis.
3. An infant with hyperthermia without a clinical cause or if is associated with toxic condition or with dehydration, on deficiency land will be suspectedUTI.
4. The most frequent contributing facts: -those that allow bacteria to invade the urinary tract, with an immature defense system at this age, were phimosis in boys, poor hygiene and PCM.
5. Urine culture is the golden method in diagnosing UTI in infants.
6. The most common microorganisms involved in the development urinary tract infections are gram negative germs: E. coli, Klepsiella, Enterobacter, and Proteus.
7. The antibiogram reveals an increased susceptibility of the germs to cephalosporin of III generation and polypeptides.
8. It requires a sustained activity to improve compliance with treatment, intimate hygiene skills, and differentiated supervision at home.

Keywords: Atypical manifestations – Infant – UTI

10. INFECTIILE CU BACTERII GRAM NEGATIVE LA SUGARII CU VARSTA SUB 12 LUNI-REZISTENTA LA ANTIBIOTICE

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Introducere: In prezent, rezistenta la antibiotice a infectiilor bacteriene Gram negative este in crestere, fiind considerata una dintre cele mai mari amenintari pentru sanatate.
Scopul studiului: determinarea rezistentei la antibiotice a infectiilor cu bacterii Gram negative la sugarii cu varsta sub 12 luni.
Pacienti si metoda: studiu retrospectiv desfasurat pe 6 ani, din care s-a selectat un lot de 87 de sugari (sub 12 luni) din anul 2014, cu uroculturi pozitive, la care s-au studiat pe langa comportamentul germenilor fata de antibiotice si alti factori care contribuie la diagnosticul si atitudinea terapeutica a infectiei.
Rezultate: distributia pe sexe este aproximativ egala, cu o varsta medie de 53,05 luni, 56% dintre acestia provenind din mediul rural. Tulpinile testate prezinta nivele ridicate de rezistenta la aminopeniciline (in special la ampicilina 77% *Escherichia coli*, 81% *Proteus mirabilis*, 100% *Klebsiella pneumoniae, Pseudomonas aeruginosa*), augumentin (37% *Proteus mirabilis*, 43% *Escherichia coli*, 64% *Pseudomonas aeruginosa*, 77% *Klebsiella pneumoniae)*, cefalosporine (Cefuroxima 31% *Escherichia coli*, 46% *Proteus mirabilis*, 69% *Klebsiella pneumoniae*, 79% *Pseudomonas aeruginosa*) si sensibilitate ridicata la carbapeneme (imipenem 98% *Klebsiella pneumoniae, Proteus mirabilis, 99% Escherichia coli;* exceptie *Pseudomonas aeruginosa*) si la colistin (99% *Pseudomonas aeruginosa, Klebsiella pneumoniae,* 100% *Escherichia coli*; exceptie: *Proteus mirabilis*).
Concluzii: multirezistenta este o problema majora a tuturor bacteriilor gram negativi iar tratamentul empiric poate sa se transforme intr-un adevarat esec cu consecinte grave, de aceea antibiograma este o atitudine indispensabila.

GRAM NEGATIVE BACTERIAL INFECTIONS IN TODDLERS YOUNGER THAN 12 MONTHS – ANTIBIOTIC RESISTANCE

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Introduction: currently, antibiotic resistance of Gram negative bacteria is on the rise, being described as one of the greatest threats to health.
Aim: assessment of the antibiotic resistance pattern of Gram negative bacteria at toddlers < 12 months of age.
Materials and methods: retrospective 6 year study from which a subset of 87 infants (below 12 months) from 2014, with positive urine cultures, have been selected to study the behavior of bacteria towards antibiotics as well as other factors that contribute to the diagnostic and therapeutic approach to the infection.
Results: patients gender distribution is approximately, with an age average of 53.05 months, 56% of them being from rural areas. Tested bacterial strains show high levels of resistance to aminopenicillins (especially ampicillin 77% *Escherichia coli*, 81% *Proteus mirabilis*, 100% *Klebsiella pneumoniae, Pseudomonas aeruginosa*), augumentin (37% *Proteus mirabilis*, 43% *Escherichia coli*, 64% *Pseudomonas aeruginosa*, 77% *Klebsiella pneumoniae)*, cephalosporins (Cefuroxima 31% *Escherichia coli*, 46% *Proteus mirabilis*, 69% *Klebsiella pneumoniae*, 79% *Pseudomonas aeruginosa*) and high sensitivity to carbapenems (imipenem - 98% *Klebsiella pneumoniae, Proteus mirabilis, 99% Escherichia coli; exception Pseudomonas aeruginosa*) and to colistin (99% *Pseudomonas aeruginosa, Klebsiella pneumoniae,* 100% *Escherichia coli*; exception: *Proteus mirabilis*).
Conclusion: the multidrug resistance is a major problem for all gram negative bacteria, and empirical treatment has the potential to become a true therapeutic failure with serious consequences, therefore performing the antibiogram is indispensable.

11. INFECTIA CU STAFILOCOC LA NOU-NASCUTI-PROBLEME DE DIAGNOSTIC SI TRATAMENT

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Introducere: Stafilococul auriu este o bacterie oportunista comensala care colonizeaza pielea si tesuturile moi. Patogenitatea este data de faptul ca poate ocoli raspunsul imun al gazdei producand atat infectii localizate cat si sistemice.
Scopul studiului: studierea colonizarii la vârstă mică cu stafilococ, dar si analiza evolutiei rezistentei la antibiotice a acestuia in culturile examinate.
Pacienți și metoda: studiu de tip retrospectiv din perioada 2012-2014, efectuat pe nou- nascuții si sugarii sub 3 luni internați in Spitalul Clinic de Copii Brasov avand diagnosticul de piodermita, infecție de bont ombilical si de sepsis. Lotul de pacienti a fost format din 255 pacienti care a corespus criteriilor de includere în studiu.

Rezultate: in lotul nostru am avut 123 fete si 132 baieti. 54.5% dintre pacienți provin din mediul urban, iar varsta medie a fost de 6.6 saptamani. Portajul nazofaringian de stafilococ este de 36.6% in perioada de nou nascut mai frecvent la cei alimentati natural.
Rezistenta la antibiotice este în crestere în anii considerati in studiul nostru cu mentinerea in platou a rezistentei la gentamicina dar cu cresterea ei la oxacilina si cefuroxima.
Concluzii: portajul nazofaringian la nou nascut nu se tratează, iar în cadrul infecțiilor considerate este imperios necesar administrarea de antibiotice su spectru cat mai restrans.

STAPHYLOCCAL INFECTION AT NEWBORN - DIAGNSOTIC AND TREATMENT CHALLENGES

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Background: Staphylococcus aureus is a comensal oportunistic bacteria that usually colonises the skin and soft tissues. Its pathogenicity is giving by the fact that it can overcome the host response determining local and systemic infections.
Aim of the study: was to assess the colonisation rate at newborns as well as to study the emergency of resistant strains in the examined cultures.
Patients and methods: retrospective study during 2012-2014 on newborns and infants under the age of 3 month, admitted at the Childrens Clinic Hospital, Braso. 255 fullfilled the diagnostic criteria and were considered in our study.
Results: there were 123 girls and 132 boys, 54.5% came from urban area and the mean age was of 6.6 weeks. Nasopharyngeal colonization was 36.6% in newborn period and was more frequently encountered at those that were breast fed. Antibiotic resistance is growing in the last years with a maintains of gentamicin and a growing pattern at oxacillin and cefuroxim.
Conclusion: nasopharyngeal colonization with staphylococcus at newborn should not be treated and if there are concerns regarding systemic infections there is a need of the corect antibiotic

CARDIOLOGIE / CARDIOLOGY

12. ENDOCARDITA INFECTIOASA SAU CUTIA PANDOREI

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Endocardita infectioasa continua sa reprezinte o importanta sursa de morbiditate si mortalitate, in ciuda progreselor de diagnostic si tratament inregistrate.
Autorii prezintaevolutia a doua cazuri de endocardita acuta infectioasa, aparute la copii cunoscuti cu malformatie cardiaca congenitala, dar care nu au fost supusi unui tratatement chirurgical corector pana la varsta de 9, respectiv 11 ani.
Primul caz, surprinde prin evolutie spectaculos favorabila, in ciuda faptului ca agentul etiologic izolat(ochrobactrum anthropi) este recunoscut ca fiind foarte agresiv si rezistent la tratamentul antibiotic. Cel de al doilea caz, endocardita cu hemoculturi negative, a avut o evolutie trenanta, cu ameliorarea lenta a tabloului clinic si parametrilor inflamatori, cu persistenta leziunilor vegetante, in ciuda nenumaratelor variante terapeutice aplicate.
Concluzie: Cazurile prezentate ilustreaza aspecte particulare ale evolutiei endocarditei infectioase si subliniaza necesitatea stabilirii deciziilor terapeutice, tinand cont de particularitatile pacientului, evolutia clinica, paraclinica si imagistica. Durata tratamentului antibiotic si toate deciziile terapeutice medicale si chirurgicale se iau cantarind atent riscurile si benefiiciile pentru a face cea mai buna alegere terapeutica.
Cuvinte cheie: endocardita, malformatii cardiace congenitale, tratament

INFECIOUS ENDOCARDITIS OR PANDORRA'S BOX

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Infective endocarditis continues to be associated with high morbidity and mortality, despite the medical and surgical therapeutic options available.

We present the clinical case of 2 children known with congenital cardiac malformation, not surgically corrected until the age of 9, respectively 11 year complicated with infectious acute endocarditis.
First case had a particularly good clinical evolution despite the fact that etiological agent was ocrobactrum antrophi, a well known agresive and resistant bacteria. The second one pose serious clinical dillema due to trenant evolution, negative hemoculture. Irrespective to various treatment regimen administrated, ecocardiographic lesions were stable with slow amelioration of clinical symptoms and inflamatorry parameters.
The present cases illustrate an unusual evolution of infective endocarditis and highlights the need for a carefully considered approach, based not only on the guidelines but also on assessment of the risks and benefits of surgical intervention, taking into account the patient's characteristics, clinical course, laboratory results and imaging studies, and. duration of antibiotic therapy, in order to decide on the best therapeutic option.
Key words: endocarditis, congenital heart disease, treatment

13. MODELE EVOLUTIVE ALE INFECTIEI STREPTOCOCICE SI RISCUL CARDIAC ACTUAL LA COPIL SI ADOLESCENT

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Autorii supun atentei, riscul complicatiilor cardiace cu evolutie subclinica de tip valvulita acuta predominant mitro-aortica in infectiile cu streptococ beta hemolitic de grup A, cu localizare rinofaringiana
Incidenta infectiilor streptococice se mentine crescuta intre 18% (endemic) si 45% (epidemic) cu localizare pe cai respiratorii superioare, pot prin mecanisme imunologice produce complicatii valvulare tip valvulita acuta agravata cu fiecare puseu. Antigenele streptococice, la organisme cu predispozitie genetica vor declansa reactii imune incrucisate mediate umoral. Similitudinea antigenic, intre proteina M, polizahardul C din membrana celulara, fac ca anticorpii specifici sa reactioneze incrucisat cu sarcolema fibrei miocardice cu glicoproteinele din structurile conjunctive ale valvelor cardiace, sinovia articulara etc. Fenomenul se declanseaza la 18- 19 zile de la debut si infectia se reacutizeaza cu fiecare puseu de infectie streptococica, ceea ce poate duce in final la constituirea unor valvulopatii de tip reumatismal. Tehnicile moderne de explorare cardiaca prin ecografie si Doppler, permite evidentierea unor regurgitatii valvulare predominent mitrale si aortice de grad I si II, care sunt evidentiate la varsta de adolescent, adult tanar. Scaderea reumatismului articular acut si a cardiopatiilor valvulare este un lucru evident, dar infectiile streptococice raman prezente, cu o incidenta crescuta.
Atunci, pe buna dreptate, se poate supune atentiei riscurile dezvoltarii unor cardite secundare, tip valvulopatie cu evolutie subclinica progresiva spre sechele cardiace valvulare, evidentiate prin ecografia Doppler cardiaca.
Cuvinte cheie: infectie streptococica, complicatie cardiaca, valvulopatie subclinica

14. ALGORITM DE DIAGNOSTIC SI TRATAMENT IN ANOMALIILE VASCULARE LA COPIL

Dr.Secheli Ionut, Dr.Secheli Marian, Dr.Draghici Isabela, Dr.Oncescu Ioan, Dr.Codrescu Mihai, Dr.Popescu Maria, Dr.Balaceanu Anca, Dr.Darasteanu Raluca, Dr.Simion Florin, Dr.Gari Iancu.
Spitalul Clinic de Urgenta pentru Copii Maria Sklodowska Curie
Impartirea anomaliilor vasculare in malformatii vasculare, hemangioame, leziuni vasculare neclasificabile sau asociate unor sindroame , ne ofera diferite optiuni de tratament in practica medicala curenta , adaptate posibilitatilor fiecarei clinici in care se prezinta astfel de pacienti. Experienta de peste 10 ani in tratarea anomaliilor vasculare ne-a permis realizarea unei clasificari anatomo-clinice pe baza careia se poate alege cu usurinta tipul de tratament: chirurgical (excizie), injectari intralezionale (Bleomocina, Ethoxiscerol, OK 432) embolizari, tratament medicamentos (Propranolol), laserterapie. Algoritmul de diagnostic si tratament poate fi un ghid in obtinerea celor mai bune rezultate in evaluarea corecta a anomaliilor vasculare.

ALGHORITHM OF DIAGNOSIS AND TREATMENT IN VASCULAR ANOMALIES IN CHILDREN

Dr.Secheli Ionut, Dr.Secheli Marian, Dr.Draghici Isabela, Dr.Oncescu Ioan, Dr.Codrescu Mihai, Dr.Popescu Maria, Dr.Balaceanu Anca, Dr.Darasteanu Raluca, Dr.Simion Florin, Dr.Gari Iancu.
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Classifcation of vascular anomalies in vascular malformations, haemangiomas, unclassified lesions, or associated with other syndroms, offer us in clinical practice different treatment for different cases of vascular anomaslies. As a result of our experiences over 10 years of treatment in vascular anomalies, we described an anatomo-clinical classification with therpy associated: surgical excisions, intralesional injections (Bleomocin, Ethoxiscerol, OK 432) , embolisations, drug therapy (Propranolol) , lasertherapy. The alghoritm we propose may be a guide in medical practice with good result in vascular anomalies therapy.

15. TULBURARI DE RITM CARDIAC CU POTENTIAL LETAL ASOCIATE UNOR SIMPTOME FRECVENTE LA COPII SI TINERI.

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Incidența simptomelor prodomale raportata la persoanele care mor subit variază statistic în funcție de metodele de studiu în jurul valorii de 50 la sută . Cele mai frecvente simptome sunt dureri în piept,lipotimia și sincopa, acestea fiind comune la tineri şi putand fi cauzate de mai multe afecțiuni cardiace și noncardiace. 20 % dintre copii prezinta o stare sincopala pana la sfarsitul adolescentei, un procent de aproximativ 10 -15 % fiind de cauza cardiaca . Sindromul WPW, TPSV, Flutterul atrial, Sindromul tahi-bradi, Fibrilatia ventriculara,BAV complet si Sindromul QT prelungit sunt anomalii de ritm cardiac ce se pot solda cu moarte subita la orice varsta. Din 3000 de decese subite de cauza cardiaca inregistrate in America pe o perioada de 7 ani, 34% s-au datorat unor tulburari de ritm cardiac nediagnosticate. In cadrul compartimentului de Cardiologie al Spitalului Clinic de Urgenta pentru copii Galati s-au monitorizat Holter Ecg in ultimul an 90 de copii ce au prezentat precordialgii,sincope, dispnee de efort, 5 dintre ei fiind diagnosticati astfel cu sindrom WPW, 3 cu TPSV, 11 cu sindrom Tahi-bradi, 2 cu BAV complet si 1 copil cu boala de nod sinusal ce a beneficiat ulterior de implantarea unui peace maker dublu cameral. 90% din pacientii monitorizati au prezentat tulburari de ritm cardiac simple sau complexe. In concluzie monitorizarea Holter Ecg a pacientilor simptomatici este extrem de utila in diagnosticarea tulburarilor de ritm cardiac cu potential letal la copii si tineri.

HEART RHYTHM DISTURBANCES POTENTIALLY FATAL ASSOCIATED WITH COMMON SYMPTOMS IN CHILDREN AND YOUTH

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Incidence of prodome symptoms reported in people who die suddenly varies statistic depending on study methods around 50 percent. The most common symptoms are chest pain, faintness and syncope, which are common in young people and can be caused by cardiac and noncardiac diseases. 20% of children shows a state of syncopal to late teens, a rate of approximately 10 to 15% were because of cardiac disturbance. WPW syndrome, TPSV, atrial flutter, Brad tachy syndrome, ventricular fibrillation, atrioventricular block and QT syndrome are abnormal heart rhythm that can lead to sudden death at any age. From 3,000 sudden cardiac deaths recorded in America for a period of 7 years, 34% were due to undiagnosed heart rhythm disorders.

In the cardiology department of the Emergency Hospital for children Galati were monitored Holter ECG in the last year 90 children who presented chest pain, syncope, exertional dyspnea, 5 of whom were diagnosed as WPW syndrome, 3 with TPSV, 11-Brad Tahi syndrome, 2 with atrioventricular complete block and 1 child with sinus node disease that subsequently received an implantable dual chamber peace maker. 90% of monitored patients had heart rhythm disturbances simple or complex. In conclusion Holter ECG monitoring in symptomatic patients is extremely useful in diagnosing heart rhythm disorders potentially lethal in children and youth.

16. CARDIOTROPHIN 1 - O NOUĂ PROVOCARE

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Cardiotrophin-1 (CT-1) este un nou membru al familiei interleukinei 6, fiind un ligand al receptorului gp-130.

Această citokină proinflamatorie s-a dovedit a fi implicată în disfuncția endotelială și hipertrofia cardiacă. Totuși semnificația clinică a CT-1 este puțin înțeleasă în principal deoarece nu există un sistem specific și sensibil aplicabil pe scară largă de testare pentru măsurarea concentrației plasmatice de-CT-1 circulant.

Nivelul seric al CT-1 se corelează cu rigiditatea arterială obiectivată prin tonometrie de apalnare. Studii recente au arătat că CT-1 s-a dovedit a fi un potent agent profibrotic pentru cord, vase și rinichi și poate determina disfuncție cardio-renală independent de valorile tensionale. Hipertrofia ventriculară stângă (HVS) este un marker independent de mortalitate la pacienţii cu hipertensiune arterială. Deşi mecanismele care contribuie la apariţia HVS sunt complexe, totuşi inflamaţia şi stressul oxidativ sunt factori determinanţi în acest proces. Deşi până în prezent, sunt puţine studii în sfera pediatrică, cercetările recente au pus în evidenţă rolul al acestui biomarker in diagnosticarea precoce a injuriei ischemice a miocardului la nou nascutii cu encefalopatie hipoxică. De asemenea nivelul seric al cardiotrophinei 1 se corelează cu riscul de a dezvolta sindrom metabolic la copii obezi.

Cuvinte cheie: cardiotrophin 1, biomarker, interleukine
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CARDIOTROPHIN 1 – A NEW CHALLENGE

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Cardiotrophin-1 (CT-1) is a new member of the interleukin-6 family, is a ligand of the receptor gp-130.
This pro-inflammatory cytokine has been shown to be involved in endothelial dysfunction and cardiac hypertrophy. However the clinical significance of CT-1 is poorly understood mainly because a specific and sensitive test applicable for measuring serum-CT-1 circulating is not available.
Serum CT-1 is correlated with arterial stiffness evaluated by the apalnation tonometry. Recent studies have shown that CT-1 has proven to be a potent profibrotic agent for heart, vessels and kidneys and cardio-renal dysfunction. Left ventricular hypertrophy (LVH) is an independent marker of mortality in patients with hypertension. Although the involved mechanisms are complex, however, inflammation and oxidative stress are determinants in this process.
Although there are few studies in the pediatric field, recent research has highlighted the role of this biomarker in the early diagnosis of myocardial ischemic injury in infants with hypoxic encephalopathy. The serum levels of cardiotrophinei 1 is correlated with the risk of developing the metabolic syndrome in obese children.
Key words: cardiotrophin-1, biomarker, interleukin
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17. APOLIPOPROTEINA A1 – UN BIOMARKER DE VIITOR

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Apolipoproteina A-I (apoA-I) umană este cea mai importantă proteină din componența lipoproteinelor cu densitate mare (HDL). Acestea dețin roluri cheie în mecanismele de prevenție a aterosclerozei. Studii importante dovedesc că protecția cardiacă atribuită acestei proteine este datorată rolului său activ în calea transportului invers al colesterolului și în prevenția disfuncției endoteliale. Se presupune că eficiența in-vivo a apoA-I depinde foarte mult de capacitatea sa de a disocia din HDL sferică dând naștere la apoA-I stabilă și săracă în lipide, care poate fi apoi rapid încărcată cu lipide ca urmare a interacțiunii cu transportorul ABCA1 de pe macrofagelor tisulare.
Obezitatea cu debut precoce în copilărie este un factor de risc major pentru obezitatea la adult și co-morbiditățile sale și se asociază cu reducerea speranței de viață. Cele mai multe dintre co-morbiditățile metabolice observate clar la adulții obezi sunt de obicei subclinice sau chiar nedetectabile la cei mai mulți tineri (prepubertari) obezi atunci când sunt utilizați biomarkerii disponibili în prezent, inclusiv adipokinele. Totuși, aceste modificări ale adipokinelor circulante asociate cu metabolismul glucidic nu s-au dovedit a fi utile în cazul detectării stadiilor incipiente ale perturbărilor homeostatice la copiii obezi.
Obezitatea infantilă trebuie luată în considerare nu numai ca un factor de risc pentru bolile cardiovasculare la vârsta adultă dar și ca o cauză de afectare subclinică a funcției cardiace în copilărie. Anomaliile metabolice care însoțesc obezitatea se asociază cu disfuncții cardiace semnificative la copil, independent de hipertensiune. Această asociere se poate datora unor factori de risc aflați în relație cu o stare inflamatorie ce poate modifica expresia unor izoforme specifice ale proteinelor serice implicate în inflamație (haptoglobina) și metabolism (apoA1).

APOLIPOPROTEIN A1 – A BIOMARKER FOR THE FUTURE

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Human apolipoprotein A-I (apoA-I) is the major protein among high density lipoproteins (HDL), associated with key functions preventing atherosclerosis. Extensive research supports that the cardio protection attributed to the protein is due to its active role in the reverse cholesterol transport pathway and in the protection against endothelial dysfunction. In order to fulfill those functions, it was speculated that the *in vivo* efficiency of apoA-I strongly depends on its ability to dissociate from spherical HDL giving rise to stable, lipid-poor apoA-I, which can be rapidly lipidated as a consequence of its interaction with cellular macrophages ATP binding cassette A1 (ABCA1) transporter. Early-onset obesity is a definite risk factor for adult obesity, its associated comorbidities and reduced life expectancy. Most of the metabolic comorbidities overtly observed in obese adults are usually subtle or even undetectable in most young (prepubertal) obese children when the currently available biomarkers, including the growing number of adipokines, are used. However, these changes in circulating adipokines related with carbohydrate metabolism have not been shown to be of value for the detection of early stages of homeostatic derangement in young obese children. Therefore, childhood obesity should be considered not only a risk factor for cardiovascular diseases in adulthood but also a cause of subclinical impairment of cardiac function in childhood. extent of increase in BMI in adolescents. We have also demonstrated how insulin resistance, the first step in obesity associated impairment of carbohydrate metabolism, enhances these changes in the degree of expression of specific isoforms of proteins related to inflammation (haptoglobin) and metabolism (ApoA1), some of them correlating with serum levels of adipokines.

18. STATUSUL NUTRITIONAL LA COPIII CU MALFORMATII CONGENITALE DE CORD

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Obiective: evaluarea starii de nutritie la copiii cu malformatii congenitale de cord (MCC).
Material si metoda. Lotul de studiu a cuprins 58 copii 0-16 ani, internati in Clinica II Pediatrie, S.C.J.U. Craiova, in perioada 2011-2014. Parametrii urmariti: tipul de MCC, varsta la diagnostic, sexul, mediul de provenienta, conditiile socio-economice, factorii de risc materni/fetali, greutatea la nastere, modul de alimentatie, parametrii antropometrici, afectiunile asociate, durata spitalizarii.
Rezultate. Prevalenta MCC a fost 0,9% in 2011, 1,8% in 2012, 0,5% in 2013, 1,2% in 2014. Cele mai multe MCC au fost la sexul masculin (71,3%), la cei proveniti din mediul rural (58,4%). 71,3% au fost MCC necianogene, 6,4% cianogene, restul MCC complexe. Distrofia/ hipotrofia ponderala a fost prezenta la 76,8%, paratrofia/supraponderea la 2,5% din cazuri (in MCC necianogene, predominand la sexul masculin, la cei din mediul rural). Alimentatia naturala a predominat la paratrofici si eutrofici. Greutatea la nastere sub 2500g a fost mai frecventa la copiii cu distrofie. 58,4% din cazurile cu distrofie au prezentat anemie si rahitism carential. 62,9% au prezentat anemia carentiala/feripriva, 7,4% retard in dezvoltarea psihomotorie, 6 copii avand sindrom Longdon-Down. S-au inregistrat 2 decese. Durata medie de spitalizare a fost 7-14 zile.
Concluzii. Prevalenta MCC a fost de 1,5%, au predominat MCC necianogene, la sexul masculin, la cei proveniti din mediul rural. Factorii favorizanti ai distrofiei au fost greutatea mica la nastere si conditiile socio-economice precare, iar ca factori determinanti avem aportul alimentar deficitar calitativ/cantitativ si diversificarea incorecta a alimentatiei.
Cuvinte cheie: status nutritional, malformatie congenitala de cord, copil

NUTRITIONAL STATUS IN CHILDREN WITH CONGENITAL HEART DEFECTS

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Objectives: the assessment of nutritional status in children with congenital heart defects (CHD).
Material and method. The group under study consisted of 58 children, aged 0-16 years, admitted in the 2nd Pediatric Clinic, Emergency County Hospital Craiova, from 2011 to 2014. Parameters to be considered: CHD type, age when diagnosed, sex, social environment, maternal/fetal risk factors, birth weight, feeding method, anthropometric parameters, associated affections, and hospitalization period.
Results. The prevalence of CHD was 0.9% in 2011, 1.8% in 2012, 0.5% in 2013 and 1.2% in 2014. The most frequent CHD were in males (71.3%), and in rural areas (58.4%). 71.3% represented non-cyanogenic CHD, 6.4% cyanogenic, the rest of them being complex CHD. Dystrophy and ponderal hypotrophy were present in 76.8%, paratrophy/overweight in 2.5% of the cases (in non-cyanogenic CHD, prevailing in males and in rural areas). The natural feeding prevailed in paratrophy and eutrophy cases. Birth weight under 2500g was more frequent in children with dystrophy. 58.4% of the cases with dystrophy presented anemia and carential rickets. 62.9% of the cases presented carential/ferriprive anemia, 7.4% retarded psychomotor development, and 6 children Langdon-Down syndrome. 2 deaths were registered. The average period of hospitalization was 7-14 days.
Conclusions. The CHD prevalence was 1.5%, more frequent being the non-cyanogenic CHD, the male cases and those coming from rural areas. Biasing factors in dystrophy were low birth weight and precarious socio-economic conditions, while

the deficit of food, both qualitatively and quantitatively, and the incorrect feeding diversification represented the determinant factors.

Keywords: nutritional status, congenital heart defects, child

19. DESCOPERIRI ECOCARDIOGRAFICE IN TULBURARILE DE RITM LA COPII

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Introducere:

Ecocardiografia poate evalua modificarile motilitatii peretilor inimii cu o precizie mai buna decat alte tehnici de investigare. Ecocardiografia M-mode utilizata impreuna cu ecocardiografia Doppler reprezinta un instrument ideal pentru diagnosticarea modificarilor aparute in activarea tulburarilor de ritm.

Material si metoda:

Am studiat 25 de copii cu tulburari de ritm si conducere internati in Clinica II Pediatrie Timisoara,in intervalul de timp 2007-2012. Acestia au fost investigati folosind electrocardiografia impreuna cu M/mode, 2/D si ecocardiografia Doppler. Grupul nostru de studiu a fost format din : 5 cazuri cu bloc de ramura dreapta, 2- cu Mobitz II tip 2, 3- cu bloc cardiac total, 5 cazuri cu ritm atrial inferior, 3- cu sindrom W-P-W, 2- cu batai ventriculare premature si 5 cazuri cu ectopie supraventriculara. Cazurile au fost evaluate inainte si in timpul tratamentului.

Concluzii: examinarea ecocardiografica in patologia pediatrica are o reala importanta in evaluarea efectelor hemodinamice ale tulburarilor de ritm si totodata pentru monitorizarea terapiei. Studiul va fi extins la un numar mai mare de cazuri incluzand si alte modificari electrice,in vederea stabilirii avantajelor hemodinamice ale diferitelor terapii si pentru a stabili toleranta la efort.

Cuvinte cheie: ecocardiografie, tulburari de ritm, copil

ECHOCARDIOGRAPHIC FINDINGS IN CHILDREN WITH ELECTRICAL ALTERATIONS

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Introduction:

Echocardiography can evaluate subtle modifications of heart's walls motility, with a better accuracy than other investigation techniques. M-mode echocardiography combined with Doppler echocardiography represents an ideal diagnosing instrument to study the functional disturbances occured with altered electrical activation.

Material and method :We studied 25 children with rhythm and conduction disturbances, hospitalized in Clinic II Pediatrics Timisoara during 2007-2012. They were investigated using electrocardiography combined with M-mode, 2/D and Doppler echocardiography. Our study group consisted of 5 cases with right bundle branch block, 2 with Mobitz II type 2 block, 3 with complete heart block, 5 cases with inferior atrial rhythm, 3 W-P-W syndromes, 2 premature ventricular beats and 5 with supraventricular ectopy. The cases were evaluated before and during the treatment.

Conclusions: echocardiographic examination in children pathology has a real importance to evaluate the hemodynamic effects of various electric disturbances and to monitor the therapy. The research will be extended on a larger number of cases, including other electric alterations to establish the hemodynamic advantages of different therapy and to establish the effort tolerance.

Keywords: ecocardiography,electric disturbances , children

20. ECOCARDIOGRAFIA IN CARDIOMIOPATIA DILATATIVA LA COPII

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Introducere :

Studiul motilitatii peretelui ventricular stang are o importanta deosebita in evaluarea cardiomiopatiei dilatative. Fiind vorba de afectarea miocardului in totalitate, este de asteptat ca toti peretii ventriculului stang sa prezinte o diminuare a contractilitatii.

Material si metoda:

Am luat in studiu un lot de 40 de pacienti diagnosticati cu cardiomiopatie dilatativa, cu varsta cuprinsa intre 5 zile si 14 ani, internati in Clinica II Pediatrie Timisoara in perioada 2002-2012. La toate cazurile am constatat o diminuare a fractiilor de ejectie si de scurtare la debutul afectiunii. S-au analizat parametri ecografici de evaluare a cardiomegaliei, insuficientei valvulare si a performantei miocardice prin masuratori in modul M si Doppler.
In concluzie, ecografia bidimensionala, M-mode si Doppler , in cardiomiopatia dilatativa, ofera informatii esentiale pentru diagnosticul pozitiv, pentru evaluarea severitatii bolii precum si pentru formularea unor indicatii terapeutice si evaluarea unui prognostic cat mai corect pentru aceasta afectiune.
Cuvinte cheie: cardiomegalie, cardiomiopatie dilatativa, copil

ECHOCARDIOGRAPHY FINDINGS IN CHILDREN WITH DILATED CARDIOMYOPATHY

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Introduction:
The study of left ventricular wall motility has a particular importance in assessing dilated cardiomyopathy. Being totally myocardial damage, it is expected that all the walls of the left ventricle to show a decrease in contractility. Material and method:
We have studied a group of 40 patients diagnosed with dilated cardiomyopathy, aged between 5 days and 14 years old, hospitalized in Pediatric Clinic II Timisoara in 2002-2012. In all cases we found a decrease in ejection fraction and shortening at the onset of disease. We analyzed the ultrasound evaluation parameters for cardiomegaly, valvular insufficiency and myocardial performance by M mode and Doppler measurements.
In conclusion, two-dimensional echocardiography, Doppler and M Mode, in dilated cardiomyopathy, provides essential information to establish the diagnosis, to assess disease severity and therapeutic indications for formulating and evaluating a more accurate prognosis for this condition.
Keywords: cardiomegaly, cardiomyopathy, children

21. ALGORITM DE DIAGNOSTIC SI TRATAMENT IN HIPERTENSIUNEA PULMONARA DIN BOLILE CONGENITALE CARDIACE LA COPIL

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Hipertensiunea pulmonară (HTP) reprezintă creșterea presiunii medii în artera pulmonară peste 25 mmHG in repaus. La copil cea mai frecventa forma de HTP este cea asociata bolilor congenitale cardiace, urmata de cea idiopatica si HTP persistenta a nou nascutului
Intre bolile congenitale cardiace se considera ca toti pacientii cu trunchi arterial comun, 50% dintre cei cu defecte septale ventriculare mari si 10-16 % dintre cei cu defecte septale atriale mari, neoperate, dezvolta in timp HTP.
Diagnosticul in HTP trebuie sa porneasca de la identificare simptomelor intalnite mai frecvent. Ecocardiografia este un examen imagistic util folosit pentru estimarea presiunilor pulmonare, insa insuficient pentru luarea unei decizii terapeutice.
Cateterismul cardiac drept (CCD) reprezinta „standardul de aur” in diagnosticul HTP. Pentru a verifica cresterea rezistentelor pulmonare, singura metoda este cea invaziva. In plus, in HTP trebuie identificati pacientii care ar putea beneficia pe termen lung de tratamentul cu blocante ale canalelor de calciu (CCB), lucru care se poate realiza prin testarea vasoreactivitatii pulmonare. CCD este indicat tuturor pacienții cu HTP pentru a confirma diagnosticul, pentru a evalua gravitatea și atunci când este luata in discutie terapia specifica medicamentoasa. O alta recomnadare de testare a vasoreactivitatii este HTP asociata bolilor congenitale cardiace, in care pacientii aflati in zona “gri” a operabilitatii (PVR index 5 -9 WU
Rp/Rs = 0.3 - 0.5).
Tratamentul in HTP este complex si trebuie adaptat etiologiei si vasoreactivitatii pulmonare. Acesta consta din aplicarea unor masuri generale, tratament de sustinere, tratament specific. Anumite forme pot beneficia de tratament complex cardiochirurgical.

DIAGNOSIS AND TREATMENT ALGORITHM IN PULMONARY HYPERTENSION IN CONGENITAL HEART DISEASES IN CHILDREN

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Pulmonary hypertension (PH) is the increase in mean pulmonary arterial pressure above 25 mmHg at rest. In children the most common form of PAH is associated with congenital heart disease, followed by the idiopathic form and the persistence of the newborn PAH.

The PAH related to unoperated congenital heart diseases is considered in all patients with common arterial trunk , 50 % of those with large ventricular septal defects and in 10-16 % of those with large atrial septal defects.
Diagnosis in PAH must start from identifying more common symptoms. Echocardiography is an extremely useful imaging examination used to estimate pulmonary pressures, but insufficient for therapeutic decision making.
Right heart catheterization (RHC) is the "gold standard" in the diagnosis of pulmonary hypertension. To check the pulmonary vascular resistance, the only method is invasive. In addition, the PAH must identify patients who might benefit from long-term treatment with calcium channel blockers (CCB), which can be achieved by testing the pulmonary vasoreactivity.
RHC is indicated for all patients with PAH to confirm the diagnosis, to assess severity and when specific drug therapy is considered. An important indication for RHC is PAH associated with congenital heart disease with patients in the "gray" zone of operability (PVR index 5 -9 WU
Rp/Rs = 0.3 - 0.5).
Treatment in PAH is complex and must be adapted to etiology and lung vasoreactivity. It consists of the application of general measures, supportive care , specific treatment. Some forms can be benefit from a complex cardiosurgical or interventional treatment (atrial septal fenestration or stenting, arterial ductus stenting, Potts shunt creation).

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22. SINCOPA LA ADOLESCENTI : DE CAUZA CARDIOLOGICA SAU NEUROLOGICA?

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Adolescent in varsta de 14 ani prezinta de la varsta de 7 ani episoade sincopale repetate, declansate dupa expunerea la soare sau efort fizic moderat . Anamnestic frecvente internari pentru aceasta patologie in serviciul de Neurologie unde a fost diagnosticat cu epilepsie idiopatica pentru care primeste tratament antiepileptic . Sub tratament frecventa crizelor sincopale creste, motiv pentru care este indrumat catre serviciul de Cardiologie. In urma monitorizarii Holter Ecg 24 ore se stabileste diagnosticul de Boala de nod Sinusal, concluzionandu-se astfel ca episoadele sincopale repetate nu reprezentau crize convulsive ci crize Adam Stokes, renuntandu-se astfel la tratamentul antiepileptic. Pacientul beneficiaza in prezent de cardiostimulare cu peacemaker dublu cameral din ianuarie 2015, moment din care nu a mai prezentat episoade sincopale.
Adolescenta in varsta de 14 ani prezinta un episod lipotimic in timpul orei de educatie fizica si sport. Anamnestic de aproximativ 2 luni acuza dispnee moderata de efort si precordialgii intermitent. Se interneaza in seviciul de Cardiologie al Spitalului Clinic de Urgenta pentru Copii “Sf Ioan”Galati unde in urma monitorizarii Holter Ecg se stabileste diagnosticul de Bloc Atrioventricular Complet, fiind indrumata ulterior catre o clinica de cardiologie interventionala in vederea implantarii de peacemaker.
In concluzie prezentam cazurile a doi adolescenti ce au prezentat episoade lipotimice ca urmare a unor tulburari de ritm cardiac cu potential letal, tulburari depistate prin monitorizare Holter Ecg.

SYNCOPE IN ADOLESCENTS : CARDIAC OR NEUROLOGICAL CAUSE?

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Adolescent aged 14 years shows from age 7 repeated syncopal episodes, triggered by exposure to sunlight or moderate exercise. In his medical history presented frequent hospitalizations for this pathology in the service of Neurology where he was diagnosed with idiopathic epilepsy and received antiepileptic treatment . Under treatment syncopal seizure frequency increases, so he is directed to the service of Cardiology. After 24 hours Holter ECG monitoring he was diagnosed with Sinus node disease, concluding this way that repeated syncopal episodes were not seizures but Adam Stokes crisis and consequently the antiepileptic treatment was stoped. The patient benefits from dual chamber pacemaker since January 2015, and he no longer presented syncopal episodes.
14-year-old girl shows a faint episode during the time of physical education and sport. Anamnesis for about two months she presented dyspnea and chest pain in moderate exercise. The services of Cardiology is hospitalized in the Emergency Hospital for Children "Saint John" Galati where ECG Holter monitoring is behind Diagnosed complete AV block subsequently being guided by an interventional cardiology clinical pacemaker implantation in order.

HEPATOLOGIE / HEPATOLOGY

23. ASPECTE ETIOLOGICE ALE ICTERULUI LA SUGAR PE O PERIOADA DE 2 ANI

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Scopul si obiectivele studiului.Icterul in primele zile de viata reprezinta o caracteristica importanta in patologia sugarului, predominand la nou-nascutii prematuri. Poate fi produs de tulburari ale productiei, captarii hepatice, conjugarii si excretiei bilirubinei.

Material si metode.Am luat in studiu un numar de 24 de copii, internati pe o perioada de 2 ani la Spitalul Municipal Filantropia.

Pacientii au fost investigati prin determinarea bilirubinei, hemoleucogramei, enzimelor hepatice, examene de urina, uroculturi, alte investigatii.

Rezultate si discutii.Pacientii studiati au provenit in majoritate din mediul urnan 14 cazuri (58,3%), au predominant cazurile de sex masculin 16 cazuri (66,6%) si s-au decelat infectii de tract urinar in 8 cazuri.

Concluzii. Infectia de tract urinar a fost un factor care a prelungit perioada de manifestare a icterului la sugar, infectia predominand cu germeni gram negativi.

Cuvinte cheie. icter, infectie de tract urinar

ETIOLOGICAL ASPECTS OF JAUNDICE IN INFANTS OVER A PERIOD OF 2 YEARS

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Introduction.Jaundice in the first days of life is an important feature in infant pathology, predominantly in premature infants. It can be produced by production disturbances, hepatic uptake, conjugation and excretion of bilirubin.

Method.I studied a total of 24 children, hospitalized for a period of two years from the Municipal Hospital Philanthropy.

Patients were investigated by determining bilirubin, blood count, liver enzymes, urinalysis, urine culture, other investigations.

Rezults and discution.The patients studied were derived mostly from rural urnan 14 cases (58.3%) were detected predominantly male 16 cases (66.6%) and urinary tract infections in 8 cases.

Conclusions. Urinary tract infection was a factor that prolonged the manifestation of jaundice in infants, the infection had been predominant with Gram negative.

Key words, Jaundice, urinary infection

24. HEPATITA COLESTATICA CU CMV LA NOU-NASCUT PREMATUR CU PATOLOGIE PERINATALA COMPLEXA

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Introducere: Spre deosebire de infectia congenitala, infectia peri- sau postnatala cu virusul citmegalic (CMV) poate avea un tablou clinic de sepsis sau de pneumonie severa, mai ales in cazul copiiilor prematuri si cu greutate mica la nastere. Hepatita determinata de CMV este frecvent intalnita la sugari, fiind asociata cu sindrom de colestaza.

Prezentare de caz: Raportam un caz de hepatita colestatica in contextul infectiei acute cu CMV la un sugar de 8 saptamani, nascut prematur, cu distrofie moderata si patologie perinatala complexa: enterocolita ulcero-necrotica, repetate crize de apnee cu bradicardie (pentru care a necesitat suport ventilator si tratament cu Dobutamina), anemie hipocroma microcitara severa si trombocitopenie moderata (pentru care a beneficiat de repetate transfuzii de masa eritrocitara si trombocitara). Asociat, a prezentat sindrom de hepatocitoliza si colestaza initial moderate, apoi agravate, insotite de hepato-splenomegalie, sindrom hepatopriv si sindrom inflamator moderat. Infectia cu CMV a fost confirmata prin anticorpi IgM anti-CMV pozitivi si ADN-CMV pozitiv.

Tratamentul a fost simptomatic si patogenetic, cu evolutie lent favorabila. In literatura de specialitate s-au descris cazuri severe de infectie care au beneficiat si de terapie antivirala (ganciclovir, valganciclovir), insa inca nu exista protocoale clare in legatura cu acest tratament in populatia pediatrica.

Concluzie: Desi infectia cu CMV la sugari este frecvent autolimitata, in cazuri severe poate fi fatala sau poate afecta pe termen lung abilitatile cognitive. Numeroase strategii de imunizare pasiva si activa sunt in dezvoltare pentru preventia infectiei congenitale, a sechelelor neurologice si a celoralte complicatii.

CHOLESTATIC CMV HEPATITIS IN A PRETERM INFANT WITH COMPLEX PERINATAL PATHOLOGY

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Introduction: Unlike the congenital infection, the perinatally or postnatally acquired infection with Cytomegalovirus (CMV) may present as a sepsis-like syndrome or severe pneumonitis, especially in the very low birth weight (VLBW) preterm infants. CMV hepatitis is relatively frequent in infancy and, in the early period, is associated with cholestasis.
Case presentation: We report a case of cholestatic hepatitis in the context of acute CMV infection in a 8-weeks old VLBW preterm infant, with moderate malnutrition and a complex perinatal pathology: necrotizing enterocolitis, recurrent episodes of apnea and bradycardia (for which he needed mechanical ventilation and Dobutamine), severe *hypochromic* microcytic anemia and moderate thrombocytopenia (for which he required numerous packed red blood cells and platelet transfusions). He also presented moderate increase of transaminases and cholestasis, then aggravated and associated with hepatosplenomegaly, impaired hepatic synthetic function and moderate acute inflammatory syndrome. The infection was confirmed by positive serological IgM-CMV antibodies and by quantitative CMV PCR assay in plasma.
Due to the symptomatic and pathogenetic treatment, his condition slowly ameliorated. There are growing evidences showing that associating this treatment with antiviral therapy (ganciclovir, valganciclovir) in severe cases was beneficial.
Conclusion: Although the CMV infection in the first year of life is often a self-limited illness, in severe cases, it may be fatal or have a negative effect on overall cognitive abilities. Numerous strategies of active and passive immunization are being developed for preventing congenital infection, neurological sequelae or any other disabilities.

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25. DIAGNOSTIC, TRATAMENT SI PROGNOSTIC IN INSUFICIENTA HEPATICA ACUTA DE CAUZA TOXICA LA COPIL

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Introducere: Insuficienta hepatica acuta (IHA) este o afectiune care se caracterizeaza prin pierderea rapida a functiei hepatice, manifestata prin icter, coagulopatie (INR>1.5) si encefalopatie hepatica, aparuta fara un context prealabil de suferinta hepatica. Managementul cazurilor de IHA la copil implica o echipa multidisciplinara (pediatru, hepatolog, nefrolog, echipa de terapie intensiva, echipa de transplant). Monitorizarea acestor cazuri trebuie realizata intr-un compartiment de terapie intensiva sau terapie intensiva intermediara. Este foarte important diagnosticul cat mai precoce si sustinerea functiilor vitale. In cele mai multe cazuri transplantul hepatic reprezinta singura optiune salvatoare, dar exista terapii specifice pentru diferite etiologii toxice, care pot reprezenta o variantă eficace în contextul urgentei (antidoturi, dializa hepatica, hemoperfuzie sau plasma-separare). Dupa infectiile virale, medicamentele (acetaminofen, valproat, albendazol) si toxicele sunt cele mai frecvente cauze de IHA la copil.
Metoda: Prezentam 3 pacienti diagnosticati si urmariti cu IHA de cauza toxica in ultimii 2 ani in clinica noastra: (1) fetita, 6 ani, cu insuficienta hepatica fulminanta dupa ingestia unei supradoze de valproat si care a prezentat evolutie favorabila dupa cateva sedinte de dializa hepatica (MARS); (2) pacienta, 10 ani, care prezinta hepatita colestatica dupa administrarea unei doze de albendazol pentru parazitoza digestive; prezenta anti-LKM a necesitat diagnosticul diferential cu hepatita autoimuna cu IHA; (3) baiat, 2 ani, cunoscut cu diagnosticul de boala granulomatoasa cronica si care a prezentat hepatita acuta toxica dupa adminstrare de paracetamol si fluconazol.
Concluzie: Am prezentat aceste cazuri pentru a ilustra dificultatile de diagnostic, optiunile terapeutice şi prognosticul IHA de cauza toxica la copil.

ACUTE LIVER FAILURE DUE TO TOXIC HEPATITIS IN CHILDREN: DIAGNOSTIC, TREATMENT AND PROGNOSTIC FACTORS

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Background: Acute liver failure (ALF) is a rare syndrome defined by rapid decline in hepatic function characterized by jaundice, coagulopathy (INR >1.5), and hepatic encephalopathy in patients with no evidence of prior liver disease. The management of ALF requires a multidisciplinary team (pediatrician, hepatologist, intensive care specialist, nephrologist, liver transplant surgeon). The most important aspect of treatment in patients with ALF is to provide good intensive care support. In most of the cases the liver transplantation may be the only saving option but, depending on the etiology, the treatment can be administration of antidotes, liver dialysis, hemoperfusion or plasma-separation. After viral hepatitis, drugs (acetaminophen, valproate and albendazole) and other intoxications are the most common etiology of ALF in children.
Methods: We present three cases of children with toxic ALF diagnosed during the last two years in our hospital: (1) female, 6 years old, with fulminant hepatic failure after ingestion of an overdose of valproate; after few session of MARS dialysis she had a spectacular clinical and biological recovery; (2) female, 10 years old, with cholestatic hepatitis after she treatment with albendazole for digestive parasitosis; she also had LKM positives, so an autoimmune hepatitis (AIH) was in her differential diagnosis; (3) male, 2 years old, with chronic granulomatous disease, developing ALF after he was treated with repeated therapeutic doses of acetaminophen and fluconazole.
Conclusion: We presented these cases to illustrate the difficulties of diagnosis, treatment options and prognosis of ALF due to toxic hepatitis in children.

26. EVALUAREA FACTORILOR DE RISC LA COPIII CU STEATOZA HEPATICĂ NON-ALCOOLICĂ

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Introducere: Boala ficatului gras non-alcoolic (NAFLD) este recunoscuta ca principala forma de boala hepatica cronica la copii, cea mai comuna cauza a cresterilor persistente inexplicabile ale transaminazelor. Material si metoda: Am efectuat un studiu pe un lot de de 57 de pacienti, monitorizati in Clinica II Pediatrie Iasi. Criteriul de includere a constat in prezenta steatozei hepatice la ultrasonografie. Criteriile de excludere au fost reprezentate de existenta cauzelor secundare de steatoza. Am urmarit prezenta factorilor de risc pentru steatoza hepatica. Rezultate: S-a observat o predominanta a steatozei hepatice la pacientii de sex masculin, 63%. Antecedentele heredo-familiale au evidentiat prezenta intrafamiliala a factorilor de risc la 44% dintre pacienti. Obezitatea s-a regasit la 59 % dintre pacienti, iar 18 % au fost supraponderali. 29% dintre pacienti au prezentat cel putin o valoare a glicemiei a jeun mai mare de 100 mg/dl. TTGO a evidentiat o scadere a tolerantei la glucoza la 3 pacienti, iar 2 au fost diagnosticati cu diabet zaharat tip II. La 58% dintre pacienti au fost prezente diverse tipuri de dislipidemie: 12% au prezentat hipertrigliceridemie, 28% hipercolesterolemie si 18% dislipidemie mixta. Hipertensiunea arteriala s-a regasit la 4 pacienti. Hipertransaminazemia initiala a fost prezenta la 68,42% dintre pacienti. Concluzii: Factorii de risc al NAFLD sunt genetici si dobanditi. NAFLD asociaza ca factori de risc modificari metabolice corelate cu obezitatea centrala, cum ar fi nivelurile crescute ale trigliceridelor, hipertensiunea arteriala, insulino-rezistenta, care cresc riscul de aparitie a diabetului zaharat de tip II si a sindromului metabolic. Cuvinte cheie:steatoza hepatica, copii, obezitate

ASSESSMENT OF RISK FACTORS IN CHILDREN WITH NON-ALCOHOLIC FATTY LIVER DISEASE

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Background: Non-alcoholic fatty liver disease (NAFLD) is recognized as the main form of chronic liver disease in children, the most common cause of unexplained persistent transaminase. Methods: We performed a study on a group of 57 patients monitored in 2nd Pediatrics Clinic Iasi. The inclusion criteria was the presence of liver steatosis on ultrasound. Exclusion criteria were the existence of secondary causes of hepatic steatosis. We assessed the risk factors for hepatic steatosis. Results: There was a predominance of fatty liver in male patients, 63%. Family history revealed the presence of intra-familial risk factors in 44% patients. Obesity was found in 59% patients and 18% were overweight. 29% of patients had at least a value of fasting glucose greater than 100 mg/dl. OGTT showed a decrease in glucose tolerance in 3 patients and 2 were diagnosed with type II diabetes. In 58% of patients were present various types of dyslipidemia: 12% had hypertriglyceridemia, 28% hypercholesterolemia and 18% mixed dyslipidemia. Hypertension was found in 4 patients. Initial hypertransaminasemia was present in 68.42% of patients. Conclusions: NAFLD risk factors are genetic and acquired. NAFLD associate as risk factors metabolic changes correlated with central obesity, such as increased levels of triglycerides, hypertension, insulin resistance, which increase the risk of type II diabetes and metabolic syndrome. Keyword:children, liver steatosis, obesity

27. COLESTAZE CU DEBUT PRECOCE

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Prezentam doi pacienti cu sindrom de colestaza instalat precoce.
Obs.1: Pacient (d.n. 27.11.2011) internat la varsta de 1 luna 14 zile pentru icter cu debut precoce, persistent in evolutie.
Datele clinice (icter, splenomegalie gr. I-II), biologice (bilirubinemie mixta asociata cu cresterea valorilor GGT si citoliza hepatica) si ecografice au sugerat diagnosticul de hepatita neonatala. VDRL (+++), TPHA (+++), la copil si mama, au precizat etiologia infectioasa a colestazei.
Asociat, pacientul prezinta: somnolenta, hernie ombilicala, fontanela posterioara > 0,5 cm, macroglosie. Rezultatul pozitiv al testului screening neonatal pentru hipotiroidismul congenital, parvenit in cursul spitalizarii, confirma suspiciunea clinica.
Obs.2: Pacienta (d.n. 16.01.2012) internata la varsta de 1 luna 10 zile pentru icter generalizat cu debut precoce (la 72 ore de la nastere), scaune decolorate si urini hipercrome, la care s-a evidentiat sindromul colestatic: hiperbilirubinemie directa, cresterea fosfatazelor alcaline si LDH, asociat cu citoliza hepatica si cresteri semnificative ale IgM CMV. Ecografia abdominala evidentiaza absenta colecistului, inlocuit cu un cordon fibros.
La 2 luni, in Spitalul Sf.Maria Iasi se practica portoenteroanastomoza Kasai, fara recuperarea in evolutie a fluxului biliar adecvat. La 9 luni se instaleaza ciroza biliara secundara. In 27.11.2012, s-a efectuat la Spitalul Bambino Gesu din Roma transplantul hepatic, cu evolutie buna pe termen lung.
Atrezia cailor biliare extrahepatice si hepatita neonatala sunt cauze frecvente de colestaza neonatala.
Diferentierea lor este fundamentala pentru ca tratamentul este diferit.
Se presupune ca etiologia lor este corelata cu o infectie antenatala.
Cuvinte cheie: colestaza, hepatita neonatala, atrezie de cai biliare

NEONATALEARLY ONSET NEONATAL CHOLESTASIS

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We are presenting two patients with early onset neonatal cholestasis syndrome.

Case 1. Patient (birth date: 27.11.2011) hospitalized at the age of one month and fourteen days, with early onset jaundice, persistent in evolution.

Clinical data (jaundice, splenomegaly I-II degree), biological data (mixed bilirubin asociated with increased values for GGT and hepatic cytolysis) and ultrasound data suggested the diagnosis of neonatal hepatitis. VDRL (+++), TPHA (+++) for mother and child, specified infectious etiology of the cholestasis.

Related, the patient presents: sleepiness, umbilical hernia, posterior fontanelle > 0,5 cm, macroglossia. The positive result of the neonatal screening test for the congenital hypothyroidism, appeared during the hospitalization, confirming the clinical suspicion.

Case 2. Patient (birth date: 16.01.2012) hospitalized at the age of one month and ten days for generalized jaundice with early onset (72 hours from birth), pale stools and dark urine, which revealed cholestatic syndrome: conjugated hyperbilirubinemia, increased alkaline phosphatase and LDH, associated with hepatic cytolysis and significant increases of the IgM CMV. The abdominal ultrasound, reveals the absence of the gallbladder, replaced with a fibrous cord.

At the age of two months, a Kasai porto enteroanastomosys is being performed without the recovery in evolution of the adequate bile flow, at St. Mary Hospital, Iasi. At the age of nine months, secondary biliary cirrhosis settles. On 27.11.2012, at Bambino Gesu Hospital, Rome, a liver transplantation was performed, with a good evolution on long term.

The extra hepatic biliary atresia and neonatal hepatitis are frequent causes of the neonatal cholestasis.

Their differentiation is fundamental, because the treatment is different.

It is supposed that their etiology is associated with an antenatal infection.

Key words**:** cholestasis, neonatal hepatitis, hepatic biliary atresia.

28. MANAGEMENTUL TERAPEUTIC AL COPILULUI CU HEPATITA CRONICA CU VHC SI MANIFESTARI EXTRAHEPATICE

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Introducere

Hepatita cronica VHC la copil ridica numeroase probleme legate de abordarea terapeutica. Managementul adecvat al acestor pacienti urmareste atat disparitia sau reducerea incarcaturii virale, cat si tratamentul manifestarilor extrahepatice.

Scopul studiului

Autorii evalueaza incidenta manifestarilor extrahepatice in infectia VHC la copil, urmarind asocierea manifestarilor extrahepatice cu alte stari patologice si evolutia naturala sub impactul terapiei specifice (antivirale) si nespecifice.

Material si metode

Studiul a cuprins 43 de copii diagnosticati cu hepatita cronica VHC si manifestari extrahepatice in Clinica II Pediatrie - Spitalul „Sf. Maria" Iasi, intre ianuarie 2009-iunie 2014.

Rezultate si discutii

Manifestarile articulare si cutanate au evoluat independent de prezenta tratamentului antiviral. Manifestarile de ordin hematologic au aparut dupa tratamentul cu Interferon la 3 (6,97%) din pacientii tratati. Anemia s-a inregistrat la 6 din pacientii tratati (13,95%), fiind asociata sau nu cu leucopenia si tombocitopenia. S-a inregistrat un caz care a evoluat spre anemie aplastica, dupa finalizarea terapiei antivirale.

Depresia a aparut la 2 (4,65%) din pacienti.

Tratamentul gastritei (cu sau fara prezenta H. pylori) a ameliorat citoliza hepatica, aceasta patologie fiind decelata cel mai frecvent. Numai la unul dintre cei 3 pacienti diagnosticati s-a putut corela prezenta gastritei si a sindromului dispeptic cu tratamentul antiviral efectuat anterior cu Interferon alfa.

Concluzii

La copil interferonul poate declansa manifestari extrahepatice cu mecanism imun. Cele hematologice sunt frecvente, reversibilitatea acestora fiind evidenta doar in cazul depresiei.Tratamentul optim al manifestarilor extrahepatice si al patologiei asociate hepatitelor cronice ramane in discutie, datorita datelor insuficiente care sa ateste eficacitatea terapiilor utilizate.

THERAPEUTIC MANAGEMENT OF EXTRAHEPATIC MANIFESTATIONS IN CHRONIC HCV INFECTION IN CHILDREN

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Introduction

HCV chronic hepatitis in children raises many problems related to the therapeutic approach. Appropriate management of these patients follows both the disappearance or reduction of viral load and treatment of extrahepatic manifestations.
Aim of the study
The authors evaluate the incidence of extrahepatic manifestations in HCV infection in children, associated with other pathological conditions and natural evolution under the impact of specific treatment (antivirals) and nonspecific therapies.
Material and methods
The study comprised 43 children diagnosed with chronic HCV hepatitis and extrahepatic manifestations in 2nd Pediatrics Clinic - "Sf. Maria " Children Hospital Iasi, Romania between January 2009 and June 2014.
Results and discussions
Joint and skin manifestations have evolved independently of antiviral therapy. Hematologic manifestations occurred after treatment with Interferon 3 (6.97%) of patients treated. Anemia was seen in 6 of the patients (13.95%), being associated or not with leukopenia and tombocitopenia. There has been one case that progressed to aplastic anemia after completing therapy. Depression occurred in 2 (4.65%) of patients.
Treatment of gastritis (with or without H. pylori) improved hepatic cytolysis, this pathology being most frequently spotted. Only one of the three patients diagnosed could correlate the presence of gastritis and dyspeptic symptoms with a previous antiviral treatment with interferon alpha.
Conclusions
In children with extrahepatic manifestations interferon can trigger immune mechanism disorders. Reversibility is obvious only in case of depression. Extrahepatic manifestations and associated pathology treatment in chronic hepatitis remains in question due to inadequate data that prove the effectiveness of the therapies used.

IMAGISTICA / MEDICAL IMAGING

29. EFECTUL FUMATULUI MATERN ASUPRA COPILULUI

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Introducere: Influența fumatului asupra stării de sănătate a copilului este cert dovedită, cu toate acestea procentul de mame fumătoare rămane considerabil, fiind crescător în țarile in curs de dezvoltare. Lucrarea de fața are ca și scop revizuirea efectelor pe care le poate avea fumatul matern asupra copilului. Incă din viata intrauterină se constată intarzaierea în crestere si dezvoltare, greutatea mica la naștere si prematuritatea fiind cele mai frecvente efecte ale fumatului asupra copilului, pe langă imaturitatea pulmonară. Frecvența patologiei respiratorii la nivelul căilor respiratorii inferioare- de tipul infecțiilor pulmonare, a wheezingului / astm, reducerea funcției pulmonare cat și afectarea căilor respiratorii superioare manifestată prin otite, adenoamigdalite este semnificativă la copilul cu mamă fumătoare. De asemenea s-a demonstrat că există un risc dublu de moarte subită la sugarii expuși fumatului pe perioada sarcinii. La nivel cognitiv si neuropsihic, este o relație directă de cauzalitate între fumatul prenatal si comportament antisocial. De asemenea, disfuncții ca reducerea abilitătilor intelectuale, deficit de atenție/ sindrom hiperkinetic sunt intalnite mai des la copiii expusi la fumat. In concluzie, fumatul pasiv, prenatal si postnatal, are o multitudine de efecte în privinta mortalității și morbidității la copii. Trebuie ca mama fumătoare sa fie conștientă de impactul acțiunii sale asupra vieții copilului și motivată în scopul renunțării la fumat.

MATERNAL SMOKING EFFECT ON THE CHILD

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Background: The influence of smoking on the health of the child is definitely proven, however the percentage of smoking mothers remains considerable and is increasing in developing countries. This paper has as purpose of reviewing the effects that maternal smoking can have on the child`s health. Ever since intrauterine life has been proven the effect on growth delay , low birth weight and prematurity are the most common effects of smoking on children, besides pulmonary immaturity. The frequency of respiratory airways infections, reccurent wheezing / asthma, reduced lung function and upper airway disease like otitis, tonsilitis is significant in children with smoking parent. It was also shown that there is a double risk of sudden

death in infants exposed to smoking during pregnancy. Regarding the cognitive and neuropsychologicalthere is a direct causal link between prenatal smoking and antisocial behavior. Also, intellectual abilities reduction dysfunction, attention deficit / hyperkinetic syndrome are more often seen in children exposed to smoking. In conclusion, secondhand smoke, prenatal and postnatal, it has a multitude of effects in terms of mortality and morbidity in children. Smoking mother should be aware of its impact on the lives of children and smoking cessation reasoned order.

30. ROLUL ECOGRAFIEI RENALE IN PERIOADA DE NOU NASCUT SI SUGAR

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Malformațiile reprezinta una dintre principalele cauze ale patologiei reno-urinare la copil, ele afectand 1% din populatia generala. Cele mai frecvent intalnite la copil sunt: refluxul vezico-ureteral (RVU), sindromul de joncțiune pielo-ureteral (SJPU), megaureterul obstructiv, valva de uretra posterioara. Scopul lucarii: Evaluarea necesitatii efectuarii ecografiei renale ca metoda de screening la nou nascut si sugar in vederea depistarii unor patologii malformative. Material si metoda: 1269 de pacienti au fost ecografiați in perioada iulie 2013 – mai 2015. Grupa de varsta a fost 0-1 an, media de 3±2.5 luni. Dintre acestia, doar 50 de pacienti (3.95%) au fost internati pentru infectie urinara. Rezultate: Patologia cea mai frecvent intalnita a fost hidronefroza congenitala: gr.I-II 125 de pacienti – 10%, gr.III-IV 25 de pacienti – 2%, iar gr.V 3 cazuri – 0.23%. Cea mai frecventa cauza de hidronefroza a fost SJPU 30 de pacienti (19.6%), urmat de RVU 15 pacienti(9.8%) si megaureterul obstructiv 10 pacienti (6.5%). Patologie malformativa: 7 cazuri de displazie renala chistica, 4 cazuri cu duplicatie pielocaliceala asociind ureterocel, 5 cazuri de agenezie renala unilaterala. Patologie tumorala: un caz de neuroblastom bilateral. Discutii: Gradele mici de hidronefroza au fost urmarite ecografic, cazurile cu peste 2 infectii urinare au efectuat cistografie, iar cele malformative si tumorale au beneficiat de imagistica superioara, fara a schimba diagnosticul stabilit ecografic. Interventie chirurgicala au necesitat 10 cazuri (0.8%). Concluzie: Ecografia renala in perioada neonatala si de sugar este importanta in stabilirea unui diagnostic complet si in monitorizarea ulterioara a acestor cazuri; este o metoda neinvaziva, accesibila, cu un raport cost/eficienta mic.

Cuvinte cheie: ecografie renala, screening

ROLE OF RENAL ULTRASOUND AS SCREENING METHOD IN NEONATES AND INFANTS

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Malformations are one of the main causes of renal pathology in children, affecting 1% of the general population. The most frequent in children are: vesicoureteral reflux (VUR), ureteropelvic junction syndrome (UPJ), obstructive megaureter and posterior urethral valve. Aim: Assessing the need to perform renal ultrasound as screening method for malformative pathology in the neonate and infant. Material and methods: Renal ultrasounds were performed on 1269 patients hospitalized during July 2013 – May 2015 . Patients were aged between 0-1 years, with an average of 3 ± 2.5 months. Of these, only 50 (3.95%) presented with urinary tract infections. Results: The most common pathology was congenital hydronephrosis I-II grade (125 patients – 10%), gr.III-IV (25 patients – 2%) and gr.V (3 cases – 0.23%). The most common cause of hydronephrosis was UPJ – 30 patients (19.6%), followed by VUR – 15 patients (9.8%) and obstructive megaureter – 10 patients (6.5%). Regarding malformations, 7 cases of renal cystic dysplasia, 4 cases of duplicated collecting system and 5 cases of unilateral renal agenesis were diagnosed. Tumoral pathology was represented by one case of bilateral neuroblastoma. Discussions: cases of low grade hydronephrosis were monitored by ultrasound, cases with more than 2 urinary tract infections underwent cystography; malformations and tumors benefited from superior imaging, without changes in diagnosis established by sonography. 10 cases (0.8%) required surgery. Conclusions: Renal ultrasound in the neonatal period and infancy is important in order to establish a complete diagnosis and subsequent monitoring of these cases. It is non-invasive, affordable, with a low cost / efficiency ratio.

Keywords: renal ultrasound, screening

31. ECOGRAFIA APENDICELUI IN URGENTELE PEDIATRICE

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Scop si obiective: studiu retrospectiv privind finalizarea unor cazuri ce au intrunit criteriile ecografice de apendicita; prezentare de imagini

Material si metode: in perioada 2011-2015 am diagnosticat ecografic, cu apendicita acuta, 19 copii care s-au prezentat in urgenta la camera de garda a Sp. Regina Maria -Baneasa; s-a utilizat ecograful Philips HD11XE, cu transductorii L12-5 si C8-5; criteriile de diagnostic au fost cele agreate de EFSUMB
Rezultate si discutii: Cei 19 copii, 12 baieti si 7 fete, au avut varste cuprinse intre 2 ani 1/2 si 14 ani. 16 au fost operati, 10 in clinica noastra si 6 in alte unitati. Ceilalti 3 au prezentat remisia criteriilor la reexaminari. Diagnosticele finale (intraoperator coroborat cu examenul anatomopatologic) au fost: apendicita catarala 2 cazuri, apendicita flegmonoasa 6 cazuri, apendicita gangrenoasa cu peritonita 3 cazuri, 1 caz de apendicita cronica, 1 caz de duplicatie apendiculara. In unul din cazurile de apendicita gangrenoasa cu peritonita localizata, tabloul clinic a fost atipic, cu prezentare pentru disurie. La 2 dintre copiii cu varstele cele mai mici, 2 ani ½, respectiv 3 ani, examenul clinic a fost nerelevant, examinarea ecografica fiind solicitata din cauza probelor biologice modificate. In 6 din cele 10 cazuri operate in clinica noastra, hemoleucograma si proteina C reactiva nu au prezentat modificari specifice.
Concluzii: Diagnosticul apendicitei si decizia operatorie pot fi dificile in cazurile cu simptome atipice si semne clinice echivoce; probele de laborator pot fi normale; ecografia aduce argumente importante diagnosticului in astfel de cazuri.
Cuvinte cheie: apendicita apendice pediatrie ecografie ultrasonografie

APPENDIX ULTRASOUND IN PEDIATRIC EMERGENCIES

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Objective: retrospective study on completion of cases that have met the sonographic criteria of appendicitis; images presentation.
Materials and Methods: Since 2011, I have diagnosed acute appendicitis, by ultrasound, in 19 children , who presented to the emergency room of Regina Maria Hospital; Philips HD11XE ultrasound system was used, with L12-5 and C8-5 transducers; the diagnostic criteria were those agreed by EFSUMB.
Results: 19 children, aged between 2 years1/2 and 14, 12 boys and 7 girls. 16 were operated, 10 in our clinic and 6 in other units. The other 3 had the remission of criteria at follow-up. Final diagnoses (intraoperatory in conjunction with pathological examination) were: 2 cases of catarrhal appendicitis, phlegmonous appendicitis 6 cases, gangrenous appendicitis with peritonitis 3 cases, 1 case of chronic appendicitis, appendicular duplication 1 case. In one case of gangrenous appendicitis with peritonitis symptoms were atypical, with dysuria at presentation. In 2 cases of small children, 2 years ½ and 3 years old, clinical examination was unreliable, ultrasound examination being requested because of altered blood tests. In 6 of the 10 cases operated in our clinic, blood count and C-reactive protein did not show specific changes.
Conclusions: The diagnosis of appendicitis and surgical decision can be difficult in cases with atypical symptoms and equivocal clinical signs. Blood tests can be normal. Ultrasound diagnosis brings important arguments in such cases.
Keywords : pediatric appendicitis appendix ultrasound sonography

32. ABCES EPIDURAL CERVICAL LA UN SUGAR DE 2 LUNI - PREZENTARE DE CAZ

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Prezentăm cazul unei fetițe în vârstă de 2 luni, ce se prezintă în clinică pentru o stare de agitație și deficit motor la nivelul membrului superior drept, cu debut relativ recent, în afebrilitate. Fără antecedente de traumatism, doar un episod de gastroenterocolită febrilă, cu aproximativ 10 zile anterior debutului simptomelor actuale. Investigațiile inițiale (puncție lombară, ecografie transfontanelară) exclud o posibilă meningoencefalită. Paraclinic însă era prezent un sindrom inflamator. S-a inițiat antibioticoterapia parenterală și s-au continuat investigațiile imagistice, luându-se în discuție o posibilă leziune cerebrală (hematom, accident vascular, tumoră) sau compresiune locală cervicală. Examenul RMN cranian și de coloană cervicală stabilesc diagnosticul de spondilodiscită C5-C6 și abces cerebral spinal epidural ce comprimă rădăcina plexului brahial drept. Localizarea abcesului, ce protruzionează anterior a făcut imposibil abordul chirurgical. S-a efectuat tratament antibiotic parenteral în asociere, timp de 6 săptămâni. Evoluția clinică a fost imediat favorabilă cu recuperare completă, iar examenele RMN în evoluție indică de asemenea o evoluție favorabilă, dar cu tendința de fuziune vertebrală, citată și în literatură ca o complicație frecventă în astfel de cazuri. Patologia descrisă la acest caz este rar întâlnită în practica pediatrică, astfel încât decizia terapeutică a fost destul de dificilă, în lipsa unui ghiduri standardizate.

SPINAL EPIDURAL ABSCESS IN A 2 MONTHS OLD GIRL.CASE REPORT

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We present the case of a 2-month-old girl, who presented at the clinic for agitation and upper right limb motor deficit with recently onset, without fever. No history of trauma, only one episode of febrile gastroenteritis, about 10 days before the actual onset of symptoms. Initial investigations (lumbar puncture, cerebral ultrasound) ruled out a possible meningoencephalitis. Paraclinically but it was now an inflammatory syndrome. Parenteral antibiotic therapy was initiated and we continued imaging investigations, taking into account a possible brain injury (hematoma, stroke, tumor) or local compressive lesion on the neck. MRI of head and neck diagnosed spondylodiscitis and C5-C6 spinal epidural abscess that compress the root of right brachial plexus. The location of the abscess, with anterior protruding made impossible the surgical approach. Parenteral antibiotic treatment was performed in combination for 6 weeks. The clinical course was favorable with complete immediately recovery and evolving MRI exams also indicate a favorable evolution, but the trend of vertebral fusion, cited also in literature as a frequent complication in such cases. The pathology described in this case is rare in pediatric practice, so that the therapeutic decision was quite difficult, in the absence of standardized guidelines.

IMUNOLOGIE / IMMUNOLOGY

33. ENCEFALITA AUTOIMUNA - POSIBILA FATETA A UNEI IMUNODEFICIENTE PRIMARE?

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Introducere: Simptomele psihiatrice pot avea cauze organice, unele dintre ele reprezentand adevarate provocari pentru medicul pediatru.
Prezentare de caz: Pacienta in varsta de 11 ani si 8 luni prezinta febra, fara asocierea unei alte simptomatologii, urmata la 2 saptamani de agitatie psihomotorie progresiva, diagnosticele variind de la depresie pana la schizofrenie. Starea generala se agraveaza cu aparitia disartriei, miscarilor diskinetice si varsaturilor. Se administreaza tratament neuroleptic, insa asocierea convulsiilor tonico-clonice in afebrilitate determina internarea, RMN-ul cerebral fiind fara modificari. Dupa 3 zile de la internare, apar febra oscilanta, miscarile coreoatetozice, hipotonia musculara si tulburarile de deglutitie, insotite de un EEG modificat, pacienta transferandu-se in sectia ATI cu suspiciunea de encefalita acuta. Investigatiile evidentiaza limfopenie si Ig M pozitiv pentru virusul herpetic si se introduce tratament cu Aciclovir, Dexametazona. Insa starea generala nu se amelioreaza si tinand cont de limfopenia severa (200/mmc) persistenta se suspicioneaza etiologia autoimuna. Investigatiile complementare exclud LES insa anticorpii anti-receptor N-metil-D-aspartat (NMDAR) sunt pozitivi confirmand diagnosticul de encefalita autoimuna. Administrarea de imunoglobulina intravenos timp de 5 zile, urmata de pulsterapie cu metilprednisolon este urmata de evolutie clinica ulterioara favorabila.
Concluzie: Encefalita autoimuna cu anticorpi anti-NMDA, entitate recent descrisa, trebuie luata in considerare in toate cazurile in care manifestarile psihiatrice sunt asociate celor organice. Studiile arata posibilitatea dezvoltarii anticorpilor anti-NMDA in cursul encefalitei herpetice, ridicandu-se suspiciunea prezentei la cazul descris a unei imunodeficiente primare specifice.
Cuvinte cheie: encefalita, NMDAR, imunodeficienta

AUTOIMMUNE ENCEPHALITIS- A POSSIBLE FACET OF A PRIMARY IMMUNODEFICIENCY?

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Introduction: Psyhiatric symptoms can have organic causes, some of them representing real challenges for the paediatrician.
Case presentation: Female patient aged 11 years and 8 months old presents with fever, without any other symptoms, followed 2 weeks later by progressive psycomotor agitation. The diagnoses varied from depression to schizophrenia. The general state alters with impaired speech, vomiting and uncontrolled movements. A neuroleptic treatement is initiated, but the assosciation of tonic clonic seizures without fever requires hospitalization and a MRI investigation, which turns out normal. After 3 days of admission, oscillating fever appears along with coreatetosis movements, muscular hypotonia, swallowing problems and a modified EEG. The patient is transferred to the Paediatric ICU with suspicion of acute encephalitis. The

investigations revealed lymphopenia and a pozitive IgM for herpes virus, hence the treatement with Acyclovir and Dexametasone is started. The outcome isn´t good and considering the severe, persistent lymphopenia (200/mmc), an autoimmune cause is suspicioned. Complementary investigations rule out SLE, but the positive result for N-methyl D-aspartate receptor antibodies confirms the diagnosis of autoimmmune encephalitis. The 5-day intravenous immunoglobulin administration, followed by methylpredinosolone pulsetherapy improved the clinical and neurological outcome of the patient.
Conclusions: NMDAR autoimmune encephalitis, a recently described disease, must be taken into consideration in all cases presenting with psyhiatric symptoms asssociated with organic manifestations. Studies show the possibility of developing NMDAR antibodies in cases of herpetic encephalitis, raising the suspicion in the presented case of a specific primary immunodeficiency.
Key words: encephalitis, NMDAR, immunodeficiency

34. PSORIAZIS VULGAR LA COPIL – PREZENTARE DE CAZ

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Copil de sex feminin, in varsta de 7 ani se interneaza prin transfer dintr-un spital judetean pentru aparitia unei eruptii cu aspect de psoriazis, in vederea precizarii diagnosticului si tratamentului. Antecedentele heredo-colaterale – o matusa paterna cu psoriazis.
La internare pacienta prezenta placi eritematoase nepruriginoase proeminente, bine delimitate, acoperite de scuame groase, albe sidefii, dispuse generalizat: la nivelul scalpului, pavilioanelor urechilor, gatului, trunchiului, membrelor, periunghial, axilar, genital. Examenul dermatologic – psoriazis vulgar in placi si placarde, diagnostic confirmat histopatologic.
Cu tratament local dermatologic evolutia a fost favorabila, pacienta prezentand initial descuamari generalizate abundente, apoi treptat reepitelizarea tegumentelor.
Am prezentat acest caz deoarece la aceasta varsta psoriazisul este rar intalnit.

PSORIASIS VULGARIS IN CHILDREN – CASE PRESENTATION

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A seven-year-old girl, with a psoriasis aspect eruption, was admitted to our clinic to decide upon the diagnosis and treatment, being transferred from a county hospital. Heredo-collateral antecedents – an aunt, related to her father, with psoriasis.
When admitted, the patient presented prominent non-pruriginous erythematous plaques, clearly marked and covered by thick, silvery-white scales, all over at the level of: scalp, earlobes, neck, torso, limbs, periungual, axilla and genital areas. The dermatological examination: psoriasis vulgaris in patches and plaques, a diagnosis also confirmed histo-pathologically.
Using a local dermatologic treatment, the evolution was favorable, the patient initially presenting abundant, generalized exfoliation, then, gradually, starting a re-epithelialization process.
We presented this case since psoriasis is rarely met at this early age.

35. PARTICULARITĂȚI CLINICE ȘI BIOLOGICE IN SINDROMUL STEVENS-JOHNSON

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Introducere. Sindromul Stevens-Johnson este o afecțiune rară, secundară unei dezordini a sistemului imun, care afectează tegumentele si mucoasele. Debutul este brusc, inprevizibil, factorii determinanți fiind medicamentoși , infecțioși, maligni sau idiopatici. Este o afecțiune gravă, potențial letală care necesită spitalizare.
Material și metodă. Autorii prezintă cazurile a 3 copii diagnosticați cu sindrom Stevens-Johnson pe baza criteriilor anamnestice, clinice și paraclinice, în clinica I Pediatrie a Spitalului Județean de Urgență Craiova în perioada 01.01.2009-30.03.2015.
Rezultate.Cazurile prezentate se caracterizează prin stare generală profund alterată, debut brusc cu edeme palpebrale sau al buzelor, hipertermie, conjunctivită și erupție eritemato-maculo-vaziculoasa aparută inițial la nivelul extremităților- obraji, palme, plante. Ulterior elementele cutanate se generalizează , apar elemente buloase care confluează, se sparg lăsând eroziuni acoperite de cruste hematice.Concomitent se produce și o afectare a mucoaselor- oculare, bucale, nazale,genitale.

Diagnosticul inițial în toate cele trei cazuri a fost de alergodermie, iar etiologia a fost medicamentoasă (Ventolin , Stratera , Sumetrolim). Vîrsta copiilor a fost între 1și 7 ani iar sex ratio F/M= 2/1. Evoluția a fost lent favorabilă (2-4 săptămăni) în toate cazurile cu restituțio ad integrum.

Concluzii

Sindromul Stevens-Johnson este o afecțiune rară care pune probleme de diagnostic diferențial în faza inițială a bolii. Evoluția este de obicei favorabilă dacă nu apar complicații.

CLINICAL AND BIOLOGICAL ASPECTS IN THE STEVENS-JOHNSON SYNDROME

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Introduction. The Stevens-Johnson syndrome is a rare condition, secondary to a disorder of the immune system, which affects the skin and mucous membranes. The onset is sudden, unpredictable, and the determining factors are drug-related, infectious, malign or idiopathic. It is a serious, life-threatening condition that requires hospitalization.

Materials and method. The authors present the cases of 3 children diagnosed with Stevens-Johnson syndrome on the basis of medical history, clinical and paraclinical criteria, in the 1st Pediatric Clinic of the County Emergency Hospital of Craiova, from 01.01.2009 to 03.30.2015.

Results. The cases presented are characterized by profoundly altered general state, sudden onset with edema of the eyelids or of the lips, hyperthermia, conjunctivitis and erythematous maculopapular rash which appeared initially on the extremities – cheeks, palms, plantar surfaces. Later on, the cutaneous elements are generalized; bullous elements conflate and break, leaving behind erosions covered by blood crusts. At the same time, there is damage to the mucous membranes – eyes, mouth, nose, genitalia.

The initial diagnosis in all three cases was allergic dermatitis, with drug-related etiology (Ventolin, Stratera, Sumetrolim). The children were aged between 1 and 7, and the sex ratio was F/M= 2/1. The evolution was slowly favorable (2-4 weeks) in all cases, with restitutio ad integrum/complete recovery to the original condition.

Conclusion**s**

The Stevens-Johnson syndrome is a rare condition that raises problems of differential diagnosis in the initial phase of the disease. The evolution is usually favorable, unless complications arise.

36. NEUTROPENIE CONGENITALA ASOCIATA CU MALFORMATIE RENALA

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Introducere. Neutropeniile congenitale reprezinta un grup heterogen de boli hematopoietice genetice determinate de diferite defecte genetice caracterizate prin scaderea numarului de neutrofile si infectii recurente si severe cu debut precoce.

Prezentare de caz. O fetita in varsta de 2 ani este internata in Clinica III Pediatrie pentru neutropenie descoperita cu ocazia investigatiilor efectuate pentru tuse convulsiva. Mama pacientei a fost diagnosticata in copilarie cu neutropenie cronica severa de etiologie necunoscuta si cu rinichi drept ectopic cu infectii urinare recurente. Nu retinem infectii recurente sau severe din istoricul de boala al fetitei. Investigatiile biologice au aratat la copil: leucocite=2700/mmc, neutrofile= 240/mmc, IgA=0,18g/l, IgG=5,6g/l, IgM=0,68g/l, limfocite B=8%, limfocite T=63% iar ecografia abdominala a evidentiat rinichi ectopic drept; mama fetitei a prezentat: leucocite=1200/mmc, neutrofile= 190/mmc, IgA=2,3g/l, IgG=8,3g/l, IgM=0,89g/l, limfocite B=7,2%, limfocite T=69,3%. Aspiratul medular a relevat mielokatexis la mama si fiica iar explorarea genetica confirma sindromul WHIM cu identificarea mutatiei c.1012 C>T p.338 R>X la nivelul exonului 2B al genei CXCR4.

Concluzii. In toate cazurile cu neutropenie cronica familiala cu mielokatexis trebuie sa ne gandim la sindromul WHIM, o imunodeficienta primara rara cu transmitere autozomal dominanta definita prin veruci, hipogamaglobulinemie, imunodeficienta si mielokatexis. Desi verucile si hipogamaglobulinemia nu este prezenta in toate cazurile, neutropenia si maduva hipercelulara sunt caracteristici obligatorii.Asocierea sindromului WHIM cu ectopia renala nu a mai fost descrisa in literatura si se pare ca aceste doua boli nu au un mecanism comun de producere.

CONGENITAL NEUTROPENIA ASSOCIATED WITH KIDNEY MALFORMATION

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Introduction. Congenital neutropenias constitute a heterogeneous group of genetic hematopoietic disorders caused by a various gene defects, which are characterized by a reduction of the absolute neutrophil count with early onset of recurrent and severe infections.
Case presentation. A 2 years old girl was admitted in the IIIrd Pediatric Clinic to establish the etiology of neutropenia occasionally discovered when she had pertussis. The patient didn't have a history of recurrent infections. The patient's mother was also diagnosed in childhood with severe chronic neutropenia of unknown etiology and ectopic right kidney with recurrent urinary tract infections but no warts until present. Laboratory investigations revealed in the child: leucocyte=2700/mmc, neutrophil= 240/mmc, IgA=0,18g/l, IgG=5,6g/l, IgM=0,68g/l, B lymphocyte=8%, T lymphocyte=63% and abdominal ultrasound had evidenced ectopic right kidney; in the mother: leucocyte=1200/mmc, neutrophil= 190/mmc, IgA=2,3g/l, IgG=8,3g/l, IgM=0,89g/l, B lymphocyte=7,2%, T lymphocyte=69,3%. The bone marrow aspiration showed myelokathexis in both patients and genetic exploration confirmed WHIM syndrome with CXCR4 mutation in 2B exon c.1012 C>T p.338 R>X.
Conclusions. In all cases with chronic familial neutropenia with myelokathexis we must think to WHIM syndrome, a rare autosomal dominant primary immunodeficiency described as warts, hypogammaglobulinemia, immunodeficiency, myelokathexis . Although verrucosis and hypogammaglobulinemia are not observed in all the cases, neutropenia and a hypercellular bone marrow are typically observed. The association with ectopic kidney wasn't described until now and probably there is no any correlation between both diseases regarding its genesis mechanism.

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37. BOALA CELIACA – UNA DIN FATETELE DEFICITULUI DE IGA

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Introducere: Deficitul de IgA, cea mai frecventa imunodeficienta primara se manifesta nu numai prin infectii recurente dar mai ales prin patologie autoimuna, de multe ori diagnosticul de imunodeficienta urmand celui de boala autoimuna.
Prezentare de caz: Pacient in varsta de 5 ani, fara istoric familial important, prezinta in noiembrie 2013 febra persistenta insotita de manifestari cutanate eritematoase, dureroase la palpare dispuse pretibial (niciodata violacee) care dispar si reapar spontan. Este tratat cu antibiotic cu remiterea febrei care insa reapare, insotita fiind de diaree cu dureri abdominale. Tranzitul intestinal se normalizeaza cu regim dietetic si tratament simptomatic insa diareea reapare periodic, soldandu-se cu stagnare ponderala. Este exclusa alergia alimentara (IgE total si specific negativ), tratata infestatia cu Giardia lamblia (examen coproparazitologic pozitiv) insa fara ameliorarea scaunelor. Tinand cont de elementele cutanate si durerile abdominale medicul de familie suspicioneaza porfiria. La internare in Clinica III Pediatrie prezinta hipotrofie ponderala, elementele pretibiale si scaune diareice alternand cu perioade de constipatie. Sunt excluse cauzele alergice, infectioase, parazitare, porfiria, iar electroforeza proteinelor serice arata hipergamaglobulinemie, imunocantitatia relevand o valoare scazuta a IgA-ului (<0.8 mg/dl), complexele imune circulante fiind pozitive. Anticorpii antitransglutaminazici au avut titru inalt (>200U/ml) iar biopsia jejunala confirma boala celiaca aceasta adaugandu-se diagnosticului final de imunodeficienta primara alaturi de cel de eritem nodos.
Concluzii: In fata oricarui caz cu diaree recurenta si/sau cronica trebuie luat in considerare dignosticul de imunodeficienta primara.
Cuvinte cheie: deficit de IgA, celiachie, copil

CELIAC DISEASE- ONE OF THE MANY SIDES OF IGA DEFICIENCY

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Introduction. IgA deficiency, the most frequent primary immunodeficiency it manifests itself not only with recurrent infections but most of the time with autoimmune pathology.
Case presentation: Five years old pacient, without significant family history, develop in November 2013 persistent fever with pretibial erythematous skin lesions (never purple) which appear and disappear spontaneously. He was treated with antibiotics but the fever reappeared and it was accompanied with abdominal pain and diarrhea which caused weight failure. There were excluded food allergy (total and specific Ig E in normal range) and the infection with Giardia lamblia (pozitive in stool exam) which was treated, but the diarrhea reappeared. Taking in account abdominal pain and skin lesions, the family doctor suspected porphyria. At the admission in our Clinic the pacient presented underweight, pretibial skin lesions and diarrhea which alternates with constipation. We have excluded porphyria, allergic, infectious and parasitic causes. The serum protein electrophoresis showed hypergammaglobulinemia.and the immunogram revealed IgA deficiency (<0.8mg/dl). The circulating immune complexes were positive. Also the antitransglutaminase antibodys were positive (>200 U/ml) and the jejunal biopsy

confirmed the diagnosis of celiac disease. The last diagnosis is added to the final diagnosis of primary immunodeficiency and erythema nodosum.
Conclusions: If we are faced with recurrent or chronic diarrhea we must also consider the possibility of a primary immunodeficiency.
Key words: IgA deficiency,celiac disease, child

38. COOPERAREA CELULARA ESTE ESENTIALA PENTRU SINTEZA DE ANTICORPI – ILUSTRARE PRINTR-UN CAZ CLINIC

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Introducere. Descoperirea cooperarii celulare B si T in sinteza anticorpica a constituit una dintre cele mai marcante descoperiri ale imunologiei. Un defect la acest nivel explica existenta cazurilor de hipogamaglobulinemie insotite de infectii tipice celulei T si nu limfocitului B.
Prezentare de caz. Un baiat in varsta de 10 luni, cu istoric familial de decese in perioada de sugar, se interneaza in Clinica III Pediatrie cu suspiciune de boala Bruton. La 6 luni a avut pneumocistoza ce a necesitat ventilatie mecanica 33 de zile. Investigatiile au aratat hemoleucograma normala IgG si IgA scazute iar IgM in limite normale. Numarul de limfocite B era normal, excluzand astfel boala Bruton, iar cel de limfocite T crescut. Au fost excluse mucoviscidoza, infectia HIV, tuberculoza. In evolutie dezvolta ulcere orale si perianale, diaree cu sange si pneumonie.Explorarile au aratat neutropenie severa-20/mmc , IgG = 0,03 g/l, IgA < 0,57g/l, IgM= 1,24 g/l. Tinand cont de sexul masculin, istoricul familial, hipogamaglobulinemia cu IgM normal si de neutropenie se suspicioneaza deficitul de CD40 ligand, diagnosticul confirmandu-se prin investigatie genetica (mutatie missense c.614 T>C in exonul 5 al genei *CD40LG),* mama fiind carrier.
Concluzii. Deficitul de CD40 ligand constituie una dintre fetele cooperarii celulare, manifestat clinic prin infectii oportuniste iar biologic prin IgG scazut cu IgM normal sau crescut, diagnosticul definitiv fiind confirmat prin biologie moleculara.
Cuvinte cheie: hipogamaglobulinemie, pneumocistoza, CD40 ligand, copil

CELLULAR COOPERATION IS ESSENTIAL FOR ANTIBODY PRODUCTION – ILLUSTRATED BY A CLINICAL CASE

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Introduction. The discovery that T cells cooperate with B cells in the induction of antibodies production marked a milestone in the study of immunology. A defect on the cooperation level also explains why we can found in clinical practice cases with hypogammaglobulinemia and infections typical for T cell deficiency and no for B cell deficiency.
Case presentation. A 10 months old male was admitted in IIIrd Pediatric Clinic Timisoara for suspicion of Bruton disease.At 6 months of age he had pneumocystosis and needed mechanical ventilation for 33 days. Explorations showed normal hemoleucogram, decreased IgG and IgA, and IgM in normal range. Number of B lymphocytes was normal, excluding so Bruton disease, and T cell subpopulations were increased. There were excluded cystic fibrosis, HIV infection, tuberculosis. In evolution he developed oral and perianal ulcers, bloody diarheea and pneumonia. Investigations revealed severe neutropenia – 20/mmc, very low IgG = 0,03 g/l, IgA < 0,57g/l, IgM= 1,24 g/l. Taking in account the male gender, family history, pneumocystosis with low IgG and normal IgM we suspected CD40ligand deficiency. Molecular analysis showed a c.614 T>C missense mutation in exon 5 of the *CD40LG* gene and mother is a carrier.
Conclusions. The deficiency of CD40LG is one face of the cellular cooperation, clinically manifested with opportunistic infection despite of the severe hypo-IgG but with normal or increased IgM , the definitive diagnosis needs molecular exploration.
Key words: hypogammaglobulinemia, pneumocystosis, CD40 ligand, child

39. DIFICULTATI DE DIAGNOSTIC INTR-UN CAZ DE LIMFOHISTIOCITOZA HEMOFAGOCITICA

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Introducere. Limfohistiocitoza hemofagocitică (HLH) reprezintă un sindrom hiperinflamator ameninţător de viaţă cauzat de o activare excesivă a limfocitelor şi macrofagelor, putand fi primara (imunodeficienţa) sau secundara (infectii, boli autoimune si maligne). Diferenţierea formelor genetice de cele dobândite este cruciala deoarece transplantul medular este o urgenţă în formele congenitale.
Prezentare de caz. Fetita in varsta de 2 ani, se transfera in Clinica III Pediatrie pentru febra prelungita, hepatosplenomegalie, scadere ponderala. Provine din sarcina nedispensarizata, fara antecedente familiale si personale semnificative. Intre varsta de 4-9 luni, a locuit la Mediterana in conditii de viata precare. Boala debuteaza in urma cu 2 luni cu febra zilnica si cresterea in volum a abdomenului. Investigatiile efectuate deceleaza pancitopenie, hipergamaglobulinemie, iar aspiratul medular si explorarile imagistice exclud patologia maligna. Urmeaza tratament antibiotic fara ameliorare si se transfera in Clinica noastra. Pe baza simptomatologiei-febra, splenomegalie, citopenie, se suspicioneaza HLH, confirmat (6 din 8 criterii) prin teste suplimentare (hipertrigliceridemie, feritina>500ng/ml, sCD25>2400Ui/ml.). Au fost excluse cauzele virale, fungice, TBC, maligne si autoimune ramanand in discutie forma primara si leishmanioza viscerala, tinand cont ca a locuit in zona Mediteraneeana. Analiza minutioasa a aspiratului medular confirma diagnosticul de leishmanioza viscerala.
Concluzii. In ciuda faptului ca Romania nu este cunoscuta ca o zona cu risc pentru leishmanioza, aceasta trebuie luata in considerare in toate cazurile de HLH, diferentierea sa de o imunodeficienta primara fiind cruciala si urgenta deoarece metodele terapeutice sunt diferite.
Cuvinte cheie: HLH, leishmania, copil

DIAGNOSTIC CHALLENGES IN A CASE OF HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS

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Introduction: Hemophagocytic lymphohistiocytosis (HLH) is a life threatening hyperinflammatory syndrome caused by excessive activation of lymphocytes and macrophages, which can be primary (immunodeficiency) or secondary (infections, autoimmune diseases and malignancy). Differentiation of the acquired from the genetic forms is crucial because bone marrow transplantation is an emergency in the congenital forms.
Case presentation. 2 year old girl, without significant personal and family history, was transferred in the IIIrd Pediatrics Clinic for prolonged fever, hepatosplenomegaly, weight loss. Between the 4 and 9 months of age she lived in precarious conditions in the Mediteranean area. The disease begins two months ago with daily fever and enlargement of the abdomen. The investigations performed detect pancytopenia, hypergammaglobulinemia, but the bone marrow aspirate and imaging exploration excluded a malignancy. Following antibiotic treatment without improvement, she's transferred in our clinic. Based on symptoms-fever, splenomegaly, cytopenia, HLH is suspected and confirmed (6 of 8 criteria) by further testing (hypertriglyceridemia, ferritin>500ng/ml, sCD25>2400 IU/ml.). We excluded viral, fungal, tuberculosis, malignant and autoimmune causes, remaining for discussion the primary form and visceral leishmaniasis, considering that she lived in the Mediterranean area. The bone marrow aspirate's thorough analysis confirmed the diagnosis of visceral leishmaniasis.
Conclusions. Despite the fact that Romania isn't known as a risk area for leishmaniasis, it should be considered in all cases of HLH, its differentiation from a primary immunodeficiency is crucial and urgent because the therapeutic methods are different.
Keywords: HLH, leishmania, child

40. BCG-ITA GENERALIZATA – UN SEMN SUGESTIV PENTRU IMUNODEFICIENTA COMBINATA SEVERA

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Introducere. Unul dintre semnele sugestive pentru o imunodeficienta primara este reactia postvaccinala severa. BCG-ita este una dintre cele mai severe complicatii intalnite la sugarii cu imunodeficienta severa (SCID) vaccinati BCG, cu repercursiune asupra evolutiei si prognosticului acestora.
Prezentare de caz. Sugar in varsta de 3 luni, sex masculin, vaccinat BCG, se interneaza cu suspiciunea de hemocromatoza ca urmare a nivelelor crescute ale transaminazelor, feritinei, infectia cu virusuri hepatitice excluzandu-se. Mama a avut doi frati care au murit in perioada de sugar prin infectie. Boala a debutat la 2 luni cu tuse si febra care nu au raspuns la tratamntul antibiotic. Examenul clinic a relevat o stare generala buna, rash la nivelul fetei si trunchiului, tuse, polipnee, fara raluri, splenomegalie, fara vreun semn clinic la locul de vaccinare BCG. Radiografia pulmonara evidentiza numeroase opacitati

paratraheale drepte. Investigatile de laborator au aratat Hb=7,4g%, trombocite=39.000/mmc, limfocite=1780/mmc (5400-7200), TGO=3119u/l, TGP=339u/l, feritina=5866µg/l , trigyceridemie=5,5 mM/l (0,7-1,7). Pe baza istoricului si manifestarilor clinice s-a exclus hemocromatoza si s-a luat in consideratie diagnosticul de limfohistiocitoza hemofagocitica. Investigatiile imunologice au evidentiat limfopenie T severa, compatibila cu diagnosticul de γc SCID, confirmat prin explorare genetica. Lavajul gastric a fost pozitiv pentru BK. Sub tratament tuberculostatic manifestarile hepatice si pulmonare s-au ameliorat, pacientul fiind transplantat medular de la donator neinrudit si compatibil.
Concluzii. Uneori semnele tipice pentru tabloul de SCID lipsesc, foarte rar acesta fiind dominat de sindromul de limfohistiocitoza hemofagocitica, in cazul nostru de infectia cu BK dupa vaccinare.
Cuvinte cheie: BCG-ita, SCID, sugar

GENERALIZED BCG-ITIS – A VERY SUGGESTIVE SIGN FOR SEVERE COMBINED IMMUNODEFICIENCY

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Introduction. One of the significant signs suggestive for a primary immunodeficiency is severe post-vaccination reaction. BCG-itis is one of the most severe diseases founded in vaccinated infants with severe primary immunodeficiencies (SCID), influencing the evolution and the prognosis of the patients.
Case presentation. Male, 3 months old, BCG vaccinated, was admitted with suspicion of hemochromatosis due to high level of transaminasis, ferritin and negative viral serology. Mother had two brothers who died in infancy due to infections. The disease started at 2 months of age with cough and fever which didn't respond to antibiotics. Clinical examination revealed a good general status, rush on the face and trunk, cough, polypneea, without crackles, splenomegaly, without any sign at the BCG vaccination site. Pulmonary Xray evidenced multiple right paratracheal opacities. Laboratory investigations showed Hb=7,4g%, Platelet=39.000/mmc, Lymphocyte=1780/mmc (5400-7200), TGO=3119u/l , TGP=339u/l, Ferritin=5866µg/l, triglyceridemia=5,5 mM/l (0,7-1,7). Hemochromatosis was excluded and was taken into discussion hemophagocytic lymphohistiocytosis. Immunological investigations performed showed severe T lymphopenia (0,1%) compatible with severe combined immunodeficiency (SCID) diagnosis, genetically confirmed as γc SCID. Gastric lavaje revealed the presence of BK. With tuberculostatic treatment, liver and pulmonary manifestations improved. The patient was unrelated matched bone marrow transplanted.
Conclusions. Sometimes the clinical and biological picture is misleading in SCID and very rare the clinical picture is dominated by hemophagocytic lymphohistiocytosis syndrome, in our case determined by BCG-itis.
Key words: BCG-it is, SCID, infant

NEFROLOGIE / NEPHROLOGY

41. MEDICATIA ANTIHIPERTENSIVA IN SINDROAMELE PROTEINURICE LA COPIL. STUDIU RETROSPECTIV IN PERIOADA 2012-2014

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INTRODUCERE: In prezent, este binecunoscut faptul ca proteinuria persistenta poate accelera progresia diferitelor nefropatii, catre insuficienta renala cronica. Efectul de renoprotectie al IECA si al BRA este bine documentat, reducerea proteinuriei prin administrarea acestor terapii fiind strans corelata cu incetinirea progresiei catre ESRD.
MATERIALE SI METODE: studiu descriptiv retrospectiv efectuat pe o perioada de trei ani, in care au fost inclusi toti pacientii cu boala renala cronica, consecinta a sindroamelor proteinurice, internati in Sectia de Nefrologie a Spitalului M.S.Curie
REZULTATE: in lotul de pacienti inclusi s-au comparat valorile proteinuriei, ale GFR, ale retentiei azotate si nivelul seric al potasiului inainte si dupa introducerea tratamentului, atat in monoterapie(IEC sau BRA) cat si prin asociere.
CONCLUZII: Rezultatele noastre sugereaza ca administrarea de blocanti ai sistemului renina-angiotensina-aldosteron, atat in monoterapie cat si in asociere, are ca efect reducerea proteinuriei, ameliorarea RFG si imbunatatirea valorilor parametrilor de retentie azotata, fara cresteri semnificative ale potasiului seric.
CUVINTE CHEIE: proteinurie, blocanti ai sistemului renina-angiotensina-aldosteron, renoprotectie

42. HIPERPARATIROIDISM SECUNDAR LA COPILUL CU BOALA CRONICA DE RINICHI - SERIE DE 5 CAZURI

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Introducere:

Tulburarea de Metabolism Fosfo-Calcic si Osos la copilul cu Boala Cronica de Rinichi asociaza complicatii majore cu morbiditate crescuta pe un organism afectat profund. La copil, nutritia, sistemul endocrin, cresterea si functile metabolice sunt esentiale pentru dezvoltarea normala a scheletului si pentru un sistem cardio-vascular competent. Odata cu scadera functiei renale, apare o deterioarare progresiva a homeostaziei minerale, cu modificari importante in hormonii circulanti, calciu si fosfor, aceste fiind obstacole importante in dezvoltarea unui os puternic.

Material si metoda:

Prezentam o serie de 5 cazuri cu hiperparatiroidism secundar sever la copii cu Boala Cronica De Rinichi in stadiu Final. Acestia au fost evaluati cardiologic, endocrinologic, imagistic, biologic si din punct de vedere al cresterii si dezvoltarii.

Rezultate:

Cardiologic, toti pacientii au prezentat hipertrofie de ventricul stang cu disfunctie diastolica importanta si cresterea grosimei intima-media la nivelul carotidei.

Endocrinologic, pacientii au prezentat intarziere in dezvoltarea sexuala cu Stadii Tanner neadecvate varstei, varsta osoasa intarziata si parathormon mult crescut.

Imagistic, am efectuat rediografii care au relevant deformari osoase importante de tip genu valgum, tal valg, matanii costale. Deasmenea 2 pacienti au prezentat fracturi de col femural cu reconsolidare deficitara.

Osteodensitometria a relevat scaderea densitatii osoase cu mai mult de doua deviatii standard la fiecare dintre acesti pacienti

Biologic, produsul Calciu - Fosfor, precum si Fosfataza Alcalina mult crescute.

Toti pacientii au prezentat intarziere importanta in dezvoltare.

Concluzii:

Desi afectarea cardiovasculara a acestor pacienti este importanta, nu au fost identificate calcificari vasculare, chiar daca unii din pacienti sunt in cadrul unui program de epurare extra-renala cu durata mai mare de zece ani.

Spre deosebire de adulti care prezinta calcificari vasculare si extrascheletare importante, copiii cu Boala Cronica Renala au afectat in special osul, un sitem in plina dezvoltare in perioada de crestere.

Mentiune : Aceasta lucrare este efectuata in cadrul Pogramului Operational Sectorial pentru Dezvoltarea Resurselor Umane (POSDRU), finantat din Fondul Social European si Guvernul Romaniei prin contractul nr. POSDRU/159 ?1.5/S/137390

Cuvinte cheie: Metabolism Fosfo-Calcic, Os, Boala Cronica de Rinichi

SECONDARY HYPERPARATHYROIDISM IN CHILDREN WITH END STAGE RENAL DISEASE - A 5 CASE-SERIES

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Introduction

Chronic Kidney Disease Metabolic Bone Disorder in children is associated with important complication that impact morbidity on an already profound affected organism.

In children, nutrition, physical activity, growth, endocrine and metabolic function is mandatory for a normal skeleton development and cardiovascular system. As kidney function decreases, a progressive deterioration in mineral homeostasis emerge, with changes in circulating levels of hormones, calcium and phosphorous, this being an important obstacles to obtain optimal bone strength and cardiovascular integrity

Methods

We present a series of 5 cases with severe secondary hyperparathyroidism in children with End Stage Renal Disease . We performed cardiological , endocrinological , imaging , biological and growth and development investigations.

Results

Cardiology: all patients had left ventricular hypertrophy with diastolic dysfunction and increased intima-media thickness of the carotid artery.

Endocrinology: the patients experienced delayed sexual development with age inappropriate Tanner stages , delayed bone age and greatly increased parathyroid hormone.

Imagistics: we conducted Radiography for each patient that revealed bone deformities (genu valgum , talus valgus) , rickets . Two patients had femoral neck fractures with poor reconsolidation .

Osteodensitometry showed bone loss of more than two standard deviations from each of these patients

Biologically: Calcium - Phosphorus product and alkaline phosphatase were greatly increased.

All patients had significant growth retardation .

Conclusions

Although cardiovascular impairment of these patients is important, vascular calcification has not been identified , even if some of the patients are in a Renal Replacement Program for more than ten years.
Unlike adults who have important vascular and extraskeletal calcifications, in children with chronic kidney disease the bone is mainly affecting, a system in full development in the chilhood .
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Key Words: Metabolic Bone Disorder, Bone, End Stage Renal Disease

43. ARTERIOPATIE UREMICA CALCIFIANTA - CAUZA RARA DE DECES LA COPILUL DIALIZAT CRONIC

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Calciphylaxis (calcific uremic arteriolopathy) is a rare, important cause of morbidity and mortality in patients with end stage renal disease (ESRD). With an incidence of 1-4% of ESRD adults, was described only in few cases in children. CASE REPORT: A 4 years old boy was in evidence of our clinic from 2 years, when he was diagnosed with ESRD, caused by an posterior urethra valve. In the evolution child developed severe osteodystrophy (phosphorus >10mg / dl, Ca x P product > 70), despite calcium carbonate therapy as phosphate-binder. The child associated constant an inflammatory syndrome, considered in context of recurrent urinary tract infections. In 08.2014 returns for monthly evaluation with dry caught, itching, scratching lesions on facies and trunk, irritability. We suspected an allergic pathology (elevated IgE, with pruritic skin lesions, persistent dry caugh), initiated therapy with Montelkast. Between September-November observed marked deterioration of nutritional status (albumin< 30g/l), heart failure, progressive respiratory failure. Child suddenly develop cardio-respiratory arrest refractory to resuscitation and die. The histopathological examination reveals diffuse dermal, lung, brain calcifications, dystrophic calcifications at left-heart, subendocardial, renal calcification and diffuse large califications in arteries, that suggest a calciphylaxis. Retrospectiv evaluation show the association of predisposants factors (refractary osteodystrophy, hypoalbuminemia, chronic inflammatory syndrome, skin lesions). This case illustrates a rare, but typical case of calciphylaxis in a boy with ESRD. The literature suggests a higher incidence in male with ESRD, secondary hyperparathyroidism, extremity and visceral organ involvement, worse prognosis. Recognition of this syndrome is mandatory, for the evolution of chid.

44. ETIOLOGIE ATIPICA DE HTA RENOVASCULARA LA COPIL

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Introducere
HTA renovasculara este principala cauza de HTA secundara la copil. De obicei valorile tensionale ridicate sunt decelate intamplator, iar etiologia vasculara se confirma in urma investigatiilor.
Prezentare de caz
Prezentam cazul unei fetite in varsta de 7ani la care s-a decelat valori ale tensiunii arteriale sistolice 180mmHg cu prilejul adenoidectomiei, firul investigatiilor conducand apoi la descoperirea malformatiei vasculare aflate la originea HTA: o artera renala supranumerara ce vascularizeaza polul superior al rinichiului stang.
In prezent valorile tensionale sunt controlate de schema terapeutica (asociere de doua antihipertensive) ramanand in observatie cardiologica si nefrologica. Tratamentul endovascular invaziv ramane de perspectiva avand in vedere varsta mica a pacientei.
Concluzie
Terapia medicamentoasa contribuie la controlul valorilor tensionale, dar rezolvarea anomaliilor vasculare este apanajul chirurgiei interventionale.

ATYPICAL ETIOLOGY FOR RENOVASCULAR HYPERTENSION IN CHILDREN

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Introduction
Renovascular hypertension is the leading cause of secondary hypertension in children. Usually high blood pressure values are detected incidentally and the vascular etiology is confirmed by investigations.
Case report
We present the case of a 7 years old girl who was detected 180mmHg systolic blood pressure values following adenoidectomy.Further investigations lead to a vascular malformation underlying the HTA: a supernumerary renal artery that vascularize the upper pole of the left kidney.
Currently BP is controlled using two antihypertensive drugs, and the patient is under cardiological and nephrological exams. Considering the patient's age, invasive endovascular treatment remains the solution for the future.
Conclusion
Drug therapy helps to control blood pressure, but interventional surgery is the prerogative for these vascular anomalies.

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45. HIPEROXALURIA PRIMARA – CAUZA DE BOALA CRONICA DE RINICHI

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Introducere
Oxaloza reprezinta depunerea sistemica de cristale de oxalat si este consecinta hiperoxaluriei primare (tip I, II, III) care asociaza BCR (boala cronica de rinichi).
Prezentare de caz
Prezentam cazul unui baiat in varsta de 11ani cu BCR stadiul terminal – hemodializa cronica pe fistula arterio-venoasa de la 9ani, pancitopenic din august 2014. AHC: un frate decedat prin IRC – Nefrocalcinoza.
In cadrul I.C.Fundeni s-a efectuat biopsie osteomedulara ce a relevat prezenta de cristale cu aspect radiar – material PAS negativ, birefringente in lumina polarizata, aspect histopatologic compatibil cu o oxaloza.
Concluzie
Solutia terapeutica pentru aceste cazuri este transplantul hepato-renal. In lipsa acestuia, afectiunea evolueaza cu acumularea de cristale (in primul rand la nivel renal) ceea ce are drept consecinta pierderea completa a functiilor metabolice, necesitand substitutia functiei renale.
Cuvinte cheie: oxaloza, rinichi, hemodializa, transplant

PRIMARY HYPEROXALURIA – CAUSE FOR CHRONIC KIDNEY DISEASE

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Introduction
Oxalosis represents systemic deposition of oxalate crystals and it is a consequence of primary hyperoxaluria (type I, II, III) which associates chronic kidney disease.
Case report
We present an 11 years old boy with end-stage renal disease (ESRD) who underwent chronic hemodialysis on arteriovenous fistula from the age of 9, and become pancytopenic in August 2014.
Familiy history reveals a brother who died at the age of 4 – diagnosed with nephrocalcinosis.
Osteomedular biopsy was practiced and it revealed the presence of crystals having radial aspect - PAS negative material with birefringence in polarized light, compatible with a histopathological aspect of oxalosis.
Conclusion
Therapeutic option for these cases is hepato-renal transplantation. Otherwise, the disease leads to crystal deposits (primarily in the kidney). The consequence is the complete loss of metabolic functions, requiring renal replacement therapy.
Keywords: oxalosis, kidneys, hemodialysis, transplantation

NEONATOLOGIE / NEONATOLOGY

46. ESTE LAPTELE MAMELOR CARE AU NĂSCUT PREMATUR, DIFERIT ?

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Introducere. Lucrarea porneşte de la premiza cunoscută a compoziţiei diferite a laptelui mamelor care au născut prematur vs. la termenul normal de gestaţie. Abordarea vizează diferenţele semnificative privind compoziţia în proteine, valoarea energetică şi raportul proteo-caloric; aceste diferenţe sunt esenţiale pentru iniţierea şi continuarea alimentaţiei enterale la nou-născuţii prematuri.
Material şi metodă. Materialul de studiu este reprezentat de un lot de 30 nou-născuţi prematuri de la ale căror mame au fost prelevate 90 de probe de lapte în diverse stadii de lactaţie precum şi stadii care au vizat o masă sau un supt. Pentru comparaţie s-au utilizat 90 de probe de lapte prelevate în condiţii similare de la un lot de 30 de mame care au născut la termenul normal de gestaţie. Toate cazurile studiate au fost spitalizate în Clinica de Obstetrică-Ginecologie şi Neonatologie a Spitalului Clinic Judeţean de Urgenţă „Pius Brînzeu" Timişoara în ultimii 2 ani. Determinarea compoziţiei probelor de lapte s-a efectuat cu ajutorul aparatului, MIRIS HMA (Human Milk Analyzer) din dotarea Clinicii, iar toate datele obţinute au fost prelucrate statistic utilizând programul de analiză statistică SPSS 16.0.
Rezultate şi discuţii. Rezultatele studiului au confirmat faptul că laptele mamelor care au născut prematur este semnificativ mai sărac în proteine, indiferent de tipul laptelui (colostru, lapte de tranziţie sau lapte matur) sau de momentul în care a fost prelevat (lapte de la început, amestec sau sfârşitul suptului) comparativ cu laptele mamelor care au născut la termen. Ca o compensare a acestui deficit, raportul proteo-caloric al laptelui mamelor de prematuri este similar celui al mamelor care au născut la termen. În privinţa proteinelor există diferenţe valorice cuprinse între 0,5g/100ml şi 1,5g/100ml în favoarea laptelui mamelor care au născut la termen. Se confirmă astfel faptul că prin naşterea prematură, neaşteptată, lactaţia mamei nu este pregătită să facă fată nevoilor de creştere foarte mari ale prematurului.
Concluzie. Organismele internaţionale de nutriţie au stabilit că necesarul de proteine, pentru iniţierea şi menţinerea alimentaţiei enterale la prematur, este foarte mare, iar pentru acoperirea sa integrală laptele mamelor care au născut prematur necesită adăugarea de fortifianţi si suplimentare proteică personalizată.

IS HUMAN MILK FROM MOTHERS WHO DELIVERED PREMATURELY, DIFFERENT?

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Introduction. The paper starts from the known statement that the milk has a different composition at mothers who gave birth prematurely vs mothers who gave birth at term. The approach aims to find a significant difference regarding protein composition, the energy and proteo-caloric ratio; these differences are essential for initiation and continuation of the enteral feeding in preterm infants.
Material and methods. The study material is represented by a group of 30 premature new-borns from whose mothers, 90 milk samples were taken at different stages of lactation and at different times during a breastfeeding session. For comparison, were used 90 milk samples taken in similar circumstances from control group of 30 mothers who gave birth at term. All cases studied were hospitalized in the Clinic of Obstetrics-Gynecology and Neonatology from the Emergency County Hospital "Pius Brînzeu" Timisoara in the last 2 years. MIRIS HMA (Human Milk Analyzer), from the Clinic, was used to determine the composition of the milk samples and all the data obtained was statistically processed using the SPSS 16.0 the statistical analysis software.
Results and discussion. Study results confirmed that the milk of mothers who gave birth prematurely is significantly lower in protein, regardless of the type of milk (colostrum, transitional and mature milk) or the moment it was taken (foremilk, mixt milk or hind milk) compared with the milk of mothers who gave birth at term. A compensation of this deficiency is the proteo-caloric ratio which is similar between mothers who gave birth prematurely and of mothers who gave birth at term. Regarding value, protein differences between 0.5g/100ml and 1.5g/100ml were noticed, favoring mothers who gave birth at term. This confirms that through premature, unexpected birth, the mothers' lactation is not ready to meet the needs of the high rate growth of prematurity.
Conclusion. International nutrition organizations has established that protein requirements, for an early initiation and continuing of enteral feeding, is very high, and to reach that requirement the milk from mothers who gave birth prematurely has to be added fortifiers and protein custom supplementation.

47. CAUZA RARA DE HIPERCALCEMIE LA SUGAR

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Socul septic complica frecvent sepsisul neonatal precoce. In rare cazuri, disfunctia circulatorie din soc, favorizeaza necroza tesutului adipos hipodermic si antreneaza complicatii metabolice severe.

Prezentam evolutia unui nou-nascut cu suferinta antepartum si sepsis neonatal precoce, care dezvolta o citosteatonecroza extinsa cu evolutie agravata de o reinfectie sistemica cu bacil piocianic.

Instalarea unui sindrom de varsaturi cu stare de agitatie, alternand cu letarige si falimentul cresterii, a impus investigarea unui sindrom metabolic. S-a descoperit o hipercalcemie majora (22 mg/dl), asociata cu alcaloza metabolica decompensata. Investigatiile paraclinice au exclus cele mai obisnuite cauze de hipercalcemie la sugar si s-a confirmat relatia dintre citosteatonecroza, hipercalcemie si alcaloza metabolica persistenta.

Raspunsul terapeutic nesatisfacator la asocierea terapiei cu diuretice de ansa (Furosemid), glucocorticoizi si hidratare parenterala, a impus completarea medicatiei cu Calcitonina si alimentatie cu formula de lapte saraca in Calciu si Vitamina D3.

Concluzie: Cazul prezentat subliniaza evolutia particulara a sepsis-ului neonatal cu citosteatonecroza, generatoare de hipercalcemie si complicata cu nefrocalcinoza asimptomatica.

A RARE CASE OF HYPERCALCEMIA IN INFANT

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Septic shock is a common complication of early neonatal sepsis. In rare cases, circulatory dysfunction in shock favors the necrosis of brown tissue, that leads to severe metabolic complications.

We present the evolution of a newborn with antepartum sufferings and early neonatal sepsis, that develops an extensive cyto steatonecrosis. The evolution is worsened by a systemic reinfection with Bacillus Piocianic.

A vomiting syndrome with agitation, lethargy and growth failure installed, that determined the investigation of a metabolic cause. A major hypercalcemia was discovered (22 mg/dl), associated with a metabolic alkalosis. Laboratory investigations have ruled out the most common causes of hypercalcemia in infants and confirmed the relationship between cyto steatonecrosis, hypercalcemia and persistent metabolic alkalosis.

Unsatisfactory therapeutic response to therapy with loop diuretics (Furosemide), glucocorticoids and parenteral hydration, required further treatment with Calcitonin and supplementing with a milk formula low in Calcium and Vitamin D.

Conclusions: The case emphasises the particular evolution of neonatal sepsis with cyto steatonecrosis, generating hypercalcemia complicated with asymptomatic nephrocalcinosis.

48. MENINGOENCEFALOCEL : ATITUDINE DIAGNOSTICA SI TERAPEUTICA

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Meningoencefalocelul reprezinta un defect de inchidere al tubului neural aparut in perioada de viata fetala, caracterizat prin protruzia, in exteriorul craniului , a tesutului cerebral, a LCR-ului . Cel mai adesea acest defect apare la nivel frontal sau occipital, in saptamana 3-4 de sarcina. Se estimeaza ca frecventa acestei patologii este de 1:10.000 nou-nascuti vii.

Nou-nascut de sex masculin, in varsta de 2 zile, provenit dintr-o sarcina cu iminenta de avort, nemonitorizata si nedispensarizata, nascut pe cale naturala, este transferat in serviciul nostru prezentand un meningoencefalocel de 5/6 cm in regiunea occipitala, acoperit de o membrana translucida cu zone hemoragice.

Nou-nascutul a fost transferat stabil cardio-respirator si hemodinamic in Clinica de Neurochirurgie Pediatrica a Spitalului Marie Curie Bucuresti. S-a intervenit chirurgical, in ziua a 4-a de viata, efectuandu-se reducerea encefalocelului. Evolutia postoperatorie este buna continuandu-se supravegherea si investigatiile.

In concluzie, este importanta diagnosticarea cat mai precoce a meningoencefalocelului, chiar in utero, fiind uneori necesara intreruperea sarcinii inca din saptamana 24 de sarcina . Dupa nastere, se intervine chirurgical de urgenta (24 ore) in cazul celor fistulizate si la 3 luni pentru cele acoperite cu tegument normal. Dupa interventie trebuie efectuat IRM cerebral pentru investigarea altor malformatii cerebrale, consult oftalmologic , recuperare medicala si dispenzarizare neurologica de durata pentru eventualele sechele neuro-psiho-motorii.

MENINGOENCEPHALOCELE: DIAGNOSTICS AND THERAPEUTICAL STANDPOINTS

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Meningoencephalocele is a neural tube defect which appears during the fetal stage, described as an abnormal sac of fluid, brain tissue, and meninges that extends through a defect in the skull. Most commonly, this defect appears in the frontal or occipital region during week 3 and 4 of pregnancy. It is estimated that the incidence of this pathology is of 1:10.000 viable newborns.

Male new-born, 2 days of age, from a pregnancy with a high miscarriage risk, unsupervised pregnancy, born naturally, was transferred into our clinic with a 5/6 cm meningoencephalocele in the occipital region, covered by a thin membrane with hemorrhagic areas.

He was transferred with stable hemodynamic and cardiorespiratory parameters to the Marie Curie Hospital, Bucharest, Pediatrics Neurosurgery Clinic. In the 4th day of life, he went into surgery for the defect repair and the reintroduction of the brain tissue into the skull. The surgery was successful with a good post-operatory evolution.

In conclusion, it is vital for the diagnostics of a meningoencephalocele to be made as soon as possible, in utero, because sometimes in the case of large defects, the pregnancy must be terminated at 24 weeks. After birth, emergency surgery must be made (first 24 hours) for the fistulised ones or at 3 months for the ones covered with normal tissue. After surgery, thorough examinations must be done: MRI for other brain abnormalities, ophtalmological consults and long term neurological follow-ups for neuro, motor and psychological sequelae.

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49. INFECTIA CU CMV, DIFICULTATI DE DIAGNOSTIC SI TRATAMENT LA NOU-NASCUT SI SUGARUL IMUNOCOMPETENT

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Introducere:Virusul citomegalic (CMV) descoperit la inceputul secolului XX are o distributie ubicuitara , fiind capabil de a infecta atât omul cât şi unele specii de animale.

Conform datelor din literatura de specialitate, prevalenţa infecţiei congenitale este de 0,2- 2,4% iar fetopatia consecutivă acesteia se situează pe primul loc în cadrul infecţiilor materno-fetale. Polimorfismul manifestărilor infecţiei congenitale se caracterizează printr-o paletă largă cu punct de plecare forme asimptomatice până la tablouri clinice variate. Majoritatea infecţiilor cu citomegalvirus nu sunt diagnosticate, acestea având tendinţa de a se reactiva intermitent, fără simptomatologie. În practică, etapele parcurse pentru diagnosticarea infecţiei cu citomegalovirus se referă la izolarea virusului şi diagnosticul imunologic. In privinta tratamentului literatura de specialitate recomandă ”abţinerea” de la instituirea terapiei antivirale în formele clinice uşoare şi moderate de infecţie cu CMV

Material si metoda: Lotul studiat a fost reprezentat de un număr de 186 de copii cu vârstă cuprinsă între 0- 18 ani, cu infecţie acută cu CMV, internaţi în Clinica II Pediatrie si Spitalului Clinic de Boli Infecţioase „Victor Babeş” Timişoara. Diagnosticul a fost confirmat serologic prin dozarea anticorpilor specifici de tip IgG şi IgM.

Rezultate şi concluzii: Din punct de vedere epidemiologic incidenţa infecţiei cu CMV a prevalat net la sugari, cu o uşoară predominanţă la sexul masculin, fără deosebire semnificativă referitor la mediul de provenienţă (rural/urban).

Concluziile studiului referitoare la tabloul clinic, investigaţiile paraclinice şi evoluţia cazurilor, determină autorii să afirme că infecţia cu CMV reprezinta o importantă problemă de sănătate publică la cuplul mamă-copil.

Cuvinte cheie: CMV, infectie, sugar

CMV INFECTION, DIFFICULTY IN DIAGNOSIS AND TREATMENT IN THE NEWBORN AND IMMUNOCOMPETENT INFANT

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Introduction :Cytomegalovirus (CMV) discovered in the early twentieth century has a ubiquitous distribution, being able to infect both human and several animal species. According to the literature, congenital infection prevalence is 0.2 to 2.4% and its subsequent fetal pathology ranks first in the maternal-fetal infections. Polymorphism manifestations of congenital infection is characterized by a wide range of asymptomatic starting point to various clinical symptoms. Most infections with CMV are not diagnosed, they tend to reactivate intermittently without symptoms. In practice, the stages for the diagnosis of CMV infection, refers to the isolation of the virus and immunological diagnosis. Regarding the treatment, scientific literature recommended "refraining" from the administering of antiviral therapy in mild and moderate clinical forms of CMV disease.

Methods: The study group was represented by a total of 186 children aged 0- 18 acutely infected with CMV, hospitalized in Pediatrics II Clinic and Infectious Diseases Hospital "Victor Babes", Timisoara. The diagnosis was confirmed by serological determination of specific antibodies IgG and IgM type.

Results and conclusions: From the epidemiological point of view, the incidence of CMV disease, prevailed in infants, with a slight predominance in males, no significant difference on the environment of origin (rural / urban).

The findings on clinical symptoms, laboratory investigations and, particularly the development of cases, determined the authors assert that, CMV infection is a major public health problem in the mother-child couple.
Keywords: CMV, infection, infant

50. COMPLICATIILE PREMATURITATII EXTREME

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Scopul studiului. Prematuritatea extrem de mica se caracterizeaza prin greutate la nastere sub 1000 g si se insoteste de multiple complicatii cu impact major asupra ratei de supravietuire.
Scopul studiului este analizarea complicatiilor perinatale ale prematuritatii extreme si incadrarea acestor prematuri intr-o categorie de nou-nascuti cu mare risc de morbditate si mortalitate neonatala.
Material si metoda. Lotul de studiu este alcatuit din 46 de prematuri cu greutatea la nastere sub 1000g, internati in Sectia Neonatologie TI si Prematuri Bega in anii 2012-2013. Este un studiu de tip transversal.
Rezultate. Repartitia in functie de sex a prematurilor ELBW: fete 52,1% si baieti 47,9%; predomina sexul feminin atat numeric, cat si prin rata supravietuirii 60,7%, comparativ cu 39,3% dintre baieti. Nasterea a fost naturala in 18 cazuri (39,1%) si prin operatie cezariana in 28 cazuri (60,9%).Greutatea medie la nastere este de 859,35+/-153g (<1000g; GN minima=400g), iar varsta gestationala medie de 26,65+/-1 saptamani (23-29 saptamani). Rata supravieturii nou-nascutilor in acest grup a fost de 60,9%.
Concluzie
Rata deceselor prin prematuritate extrema este mare, fiind implicate cel mai frecvent complicatii respiratorii, infectioase, neurologice.
Cuvinte cheie: complicatii, prematuritate extrema, greutate la nastere

THE COMPLICATIONS OF THE EXTREME PREMATURITY

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Introduction. The extremely low birth weight (ELBW) infant is weighing under 1,000g at birth.
This study aimed to analyse the perinatal complications of extreme prematurity, leading to increased infant morbidity and mortality risks for extremely low birth weight babies.
Material and method. The study group comprises 46 ELBW newborns, birth weight (BW) <1,000g, admitted to the Neonatal Intensive Care Unit of *Bega* Maternity Hospital Timisoara, Romania in the period of 01.01.2012-31.12.2013. This is a transversal (retrospective observational) study.
Results. The study group comprises 46 ELBW infants, treated in the Neonatal Intensive Care Unit of *Bega* Maternity Hospital Timisoara within two years.
The gender distribution of ELBW preterms was as follows: 52.1% girls and 47.9% boys; the female gender prevails both in number and survival rate: 60.7% girls, compared with 39.3% boys.
The mode of delivery was vaginal in 39.1% and caesarean section in 60.9% of the cases.
The mean birth weight was 859.35+/-153g (<1,000g; lowest BW=400g), with the mean gestational age of 26.65+/-1 week (23-29 weeks).
Survival rate of ELBW preterm babies in the study group was 60.9%.
Conclusion
The rate of deaths from extreme prematurity is high, most commonly complications involved :respirator, infectious, neurological.
Keywords: complications, extreme prematurity, birth weight

51. CORELATII INTRE FACTORII DE RISC ANTENATALI AI PREMATURITATII SI COMPLICATIILE PREMATURITATII (SINDROMUL DE DETRESA RESPIRATORIE PRIN DEFICIT DE SURFACTANT)

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Scopul studiului. Factorii de risc antenatali sunt asociati cu incidenta crescuta a prematuritatii (8-10% din totalul nasterilor; până la 25 % din nasteri, în tarile în curs de dezvoltare).
Factorii de risc ai prematuritatii si prezentasindromului de detresa respiratorie prin deficit de surfactant sunt in corelatie.

Material si metoda. Lotul de studiu cuprinde 46 de prematuri cu greutate sub 1000 g (reprezentand 9,5% din totalul de 484 de prematuri), tratati în Clinica Neonatologie a Bega, în perioada 2012-2013.Datele din foile de observatie au fost prelucrare, rezultatele obtinute permitand incadrarea factorilor de risc ai prematurităţii în categorii.
Rezultate. Cel mai frecvent sunt intalniti:1)factorii socio-economici: statutul socio-economic precar 38%, fumatul 21,4%; 2) factorii de risc materni ai prematurităţii extreme: avorturi în atecedente 52%, anemia gravidică 40%, HTAIS 19%, sindromul Hellp 16,7%; 3) factorii de risc obstetricali: infectii intraamniotice 54,7%, circulară de cordon ombilical 16,6%, asezarea transversă 15,5%; 4) factorii de risc fetali:prematuritatea extremă 100%.
SDR prin deficit de surfactant este prezenta in proportiile: forma usoara 3,6%, forma moderata 8,6%, forma severa 80,3%.
Concluzii. Factorii se risc ai prematuritatii se asociaza cu o incidenta crescuta a prematuritatii si a SDR prin deficit de surfactant. Cunoasterea factorilor de risc ai prematurităţii impune aplicarea metodelor de preventie a nasterilor premature.
Limitastudiuluiestenumarulmic de indiviziinclusi in lot.
Cuvinte cheie: detresa respiratorie, factori de risc ai prematuritatii, prematuritate.

THE CORRELATIONS BETWEEN THE ANTENATAL RISK FACTORS OF PREMATURITY AND THE COMPLICATIONS OF THE PREMATURITY (THE SURFACTANT DEFICIENCY RESPIRATORY DISTRESS SYNDROME)

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Aim of study. The antenatal risk factors are associated with increased incidence of prematurity (8-10% of all births, up to 25% of births in developing countries).The risk factors of prematurity and the presence of the surfactant deficiency respiratory distress syndrome are correlated.
Material and method. The study group consists of 46 premature infants weigh ingless than 1000g (representing 9.5% of the total of 484 premature infants), treated in the Clinic of Neonatology Bega, during 2012-2013. Data from case report forms were processing, allowing classification results prematurity risk factors in categories.
*Results.*The most frequently encounter are: socio-economical conditions 38%, smoking 21.4%; maternal risk factors of extreme prematurity-abortions in 52%, gravidarum anemia 40%, HTAIS19%, 16.7% Hellp syndrome; obstetrical risk factors-intraamniotically infections 54.7%, circular umbilical cord 16,6%, transverse settlement15.5%; the fetal risk factors-extreme prematurity100%. RDSis present in proportions: mild 3.6%, 8.6% as moderate, severe form 80.3%.
Limit of the study is the small number of individuals included in the lot.
Keywords: prematurity, respiratory distress, risc factors for prematurity.

NEUROLOGIE SI PSIHIATRIE INFANTILĂ / CHILD NEUROLOGY AND PSYHIACTRY

52. UN CAZ DE PATOLOGIE GASTROENTEROLOGICA TRIMIS PE SECTIA DE NEUROLOGIE PEDIATRICA

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Introducere

O pacienta in varsta de 1 an 9 luni, cu dezvoltare neuropsihomotorie normala, al carei computer tomograf (CT) cerebral a relevat atrofie temporo-parietala bilaterala, a fost trimisa catre departamentul nostrum pentru episoade recurente de varsaturi alimentare.

Materiale si Metode

Copilul a fost evaluat pe sectia noastra din punct de vedere clinic si prin inregistrari electroencegalografice (EEG) de veghe si de somn. La o internare ulterioara, a fost evaluata si in departamentul de gastroenterologie, prin analize sangvine si din scaun, ecografie abdominala si endoscopie digestive superioara cu biopsie de mucoasa gastrica.

Rezultate si discutii

Examenul neurologic al copilului fost normal. Avand in vedere manifestarile clinice si aspectul CT cerebral, s-au luat in discutie crizele epileptice de origine temporala insa inregistrarile EEG, atat in veghe cat si in somn, au fost normale. O alta

ipoteza a fost un sindrom de varsaturi ciclice, acesta fiind insa un diagnostic de excludere, nu a putut fi luat in considerare in lipsa unei evaluari amanuntite gastroenterologice. Endoscopia digestive superioara a relevat un aspect de gastrita, ce a fost dovedita ulterior a fi cauzata de o infectie cronica cu Helicobacter Pylori. Manifestarile clinice ale copilului s-au remis dupa tratamentul corect si eradicarea infectiei.

Concluzii

Cazul prezentat ilustreaza importanta unei echipe multidisciplinare in evaluarea pacientilor pediatrici, avand in vedere diversitatea etiologiilor ce stau la baza manifestarilor clinice intalnite.

Cuvinte cheie: episoade de varsaturi, EEG, copil

A CASE OF GASTROENTEROLOGICAL DISEASE REFERRED TO THE NEUROLOGY DEPARTMENT

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Background

A girl aged one year and 9 months, with normal neuropsychomotor development, whose previous cerebral computed tomography (CT) showed bilateral temporo - parietal atrophy, was referred to our Department for recurrent stereotyped episodes of vomiting.

Materials and Methods

We evaluated the child clinically and performed electroencephalograms (EEGs) during both wake and sleep. During a later admission she was investigated in the Gastroenterology Department, undergoing extensive blood and stool workup, abdominal ultrasound and upper gastrointestinal endoscopy with gastric mucosal biopsy.

Results and Discussions

The neurologic examination of the patient was normal. Given the clinical manifestations and the cerebral CT aspect, we took into consideration the possibility of epileptic seizures with a temporal origin. The EEG recordings, in wakefulness as well as in sleep were normal. Another hypothesis was that of a cycling vomiting syndrome but that was an exclusion diagnosis, which could not be taken into consideration in the absence of a thorough gastroenterological evaluation. The upper gastrointestinal endoscopy revealed an aspect of gastritis, which was later confirmed to be caused by a chronic infection with H. Pylori. The clinical manifestations subsided under correct treatment.

Conclusions

The case illustrates the importance of a multidisciplinary approach in dealing with pediatric patients, since each symptom can be put forward by a multitude of different etiologies, under different pediatric disciplines.

Key words: episodes of vomiting, EEG, child

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53. MIELITA TRANSVERSA DEBUT ATIPIC LA ADOLESCENTA

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Obiective: evidentierea diagnosticului corect al unei mielite transverse cu debut atipic si instituirea prompta si corecta a tratamentului corespunzator.

Material si metode: Prezentam cazul unui adolescent venit la camera de garda a CPU din Spitalul Clinic de Urgenta “Sf Ioan” Galati. Pacientul s-a prezentat cu paraplegie flasca , insotita de glob vezical, precedata de 3 saptamani de deficit monocrural si dureri monocrurale, urmata in ziua prezentarii de deficit complet pe ambele membre. Debutul simptomatologiei nervoase a fost precedat si de o infectie de tract respirator superior.

Rezultate si concluzii: Este necesara diagnosticarea corecta, in primul rand, pe baza tabloului clinic, avand in vedere antecedentele infectioase aparute, desfasurarea spatio-temporala a evenimentelor , manifestate ca o viroza pulmonara, antecedente insotite apoi de aparitia si accentuarea starii de oboseala generalizata, de dureri la nivelul membrelor, la care se adauga deficitele motorii, senzitive, sfincteriene specifice afectarii mielitei transverse. Al doilea criteriu de diagnosticare a mielitei transverse il reprezinta investigatiile paraclinice: testele sangvine, examenul LCR si IRM, acesta fiind deosebit de important în stabilirea diagnosticului.

Cuvinte cheie: mielita,subacut,rara,tratament

TRANSVERSE MYELITIS, ATYPICAL TEENAGE DEBUT

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Objectives highlighting transverse myelitis correct diagnosis of an atypical onset and establishment of prompt and accurate treatment accordingly.
Material and Methods: We present the case of a teenager came to the emergency room of a hospital emergency CPU "Saint John" Galati. This patient presented with flaccid paraplegia, accompanied by bladder globe, preceded by three weeks of pain and monocrurale monocrural deficit, followed on the day of the full deficit on both States. Onset of symptoms was preceded nerve and upper respiratory tract infection.
Results and conclusions: correct diagnosis is necessary, firstly, based on the clinical picture, considering emerging infectious history, spatio-temporal unfolding of events, manifested as a pulmonary virosis, accompanied then by the emergence history and emphasizing generalized fatigue, pain in the limbs, plus motor deficits, sensitive, specific sphincter damage transverse mellitus. The second criterion is the transverse mellitus diagnostic laboratory investigations: blood tests, MRI and CSF examination, this is particularly important in diagnosis.
Keywords: myelitis, subacute rare treatment

54. THE CAUCASIAN CHALK CIRCLE: AGGRESSIVE PARENTS AND DIVIDED CHILDREN

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INTRODUCTION: the abstract title artistic-literary reference (theatrical work by B. Brecht of 1944) properly introduces the problem underlying the "Parental Alienation Syndrome" or PAS. This syndrome was codified in 1985 by R. Gardner, forensic psychiatrist at Columbia University, and since then it represents more cross that delight for families, courts, psychologists and civil society.
DISCUSSION: the most significant scientific literature (legal, judiciary, pediatric, psycho-sociological framework) and main legislative references (l. 54/2006 treating shared custody and its proposed revisions) were reviewed. Moreover was examined the issue regarding the gold standard for the child placement (art. 403 of C.p.c and its proposed revisions), after separation or divorce, between familiar and extra-familiar structures (community, foster home), considering child right to both parents and with regard to practice achievable uses. The analysis method and the interpretation key used reflect the need for a “new style” of hermeneutical, heuristic and anthropologic approach to the problem, respecting the reasons of objectivity related to incandescence of matter. This approach would flow from the fundamental function of “advocacy” that the modern childhood culture assigns to the pediatrician. This function, that passes to a child-centered global vision postulated in documents, declarations and statements (see art.3 of Conv. on the Child Rights), still remains in fact mortified and weakened in various parts of the world.
CONCLUSIONS: the PAS still provides a painful demonstration that its adult-centered myopic interpretation (started from the same R. Gardner) continues to produce immediate and long-lasting damages for the involved actors from the child, first victim, to the alienated parent. Failure of the preventive, clinical, judicial and rehabilitative actions requires a reinterpretation of the phenomenon by childhood professionals in the various areas, lawyers and magistrates engaged in conflicting separations to imagine and create, with the indispensable appropriate legislation aid, innovative new ways of real protection to victims of PAS.

GASTROENTEROLOGIE/ GASTROENTEROLOGY

55. DIAREEA ACUTA INFECTIOASA LA COPII. POSIBILITATI DIAGNOSTICE

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Diareea acuta infectioasa (DAI) este determinata de multiplii agenti etiologici. Evaluarea etiologica se bazeaza pe teste de laborator "de rutina" sau efectuate "la cerere". Obiective. Evaluarea rezultatelor testelor efectuate la copiii cu DAI timp de 1 an (august 2013– iulie 2014). Metode. Autorii au considerat ca evaluare "de rutina" coproculturile pentru *Salmonella* spp., *Shigella* spp. si *Campylobacter* spp. S-au efectuat "la cerere": coprocultura pentru *E.Coli* enteropatogen (ECEP), *E.Coli* enterohemoragic (ECEH), *Yersinia enterocolitica* si detectarea antigenelor Rotavirus/ Adenovirus. Medii cultura utilizate: Hectoen-enteric, bulion Selenit, Sorbitol MacConkey, agar CIN, CCDA modificat. Alte teste: latex-aglutinare, teste biochimice, aglutinare cu ser anti-"O", coloratie Gram, carduri Vitek de identificare, teste imunocromatografice (antigene virale). Criterii excludere: culturile de control pozitive. Rezultate. S-au efectuat 1998 coproculturi "de rutina", 196 "la cerere" pentru *Yersinia*, 108 pentru ECEP, 24 pentru ECEH si 1303 teste rotavirus/adenovirus. 185 (9,25%) coproculturi au fost pozitive: 89 pentru *Campylobacter*, 67 pentru *Salmonella*, 13 pentru *Shigella*, 9 pentru *Yersinia*, 7 pentru ECEP. 294 teste virale (22,5%) au fost pozitive: rotavirus 80%, 20% adenovirus. Concluzii. 1.Etiologia virala a fost cea mai frecventa, cu 2 varfuri de incidenta (mai si decembrie). 2.*Campylobacter* a fost cea mai frecventa etiologie bacteriana urmata de *Salmonella*; 3.Jumatate din tulpinile *Yersinia* au fost izolate fara "cerere speciala" din partea clinicianului, pe perioada verii, sugerand utilitatea folosirii mediului CIN in examinarea "de rutina"; 4.Autorii nu au identificat tulpini ECEH la pacientii cu sindrom hemolitico-uremic, justificand reevaluarea metodei; 5.Tulpinile ECEP ar trebui confirmate utilizand teste moleculare.

Cuvinte cheie: diaree acuta infectioasa, coprocultura, copil.

INFECTIOUS ACUTE DIARRHEA IN CHILDREN. DIAGNOSIS POSSIBILITIES

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Infectious acute diarrhea (IAD) is due to multiple etiological agents. Etiology identification is based on laboratory tests accomplished by routine assay or special request. Objectives. To evaluate tests' results for IAD diagnosis in children during 12 month-period (August 2013– July 2014). Methods. The authors considered as "routine evaluation" stool culture for *Salmonella* spp., *Shigella* spp. and *Campylobacter* spp. There were performed, at "special request": stool culture for enteropathogenic *E.Coli* (EPEC), enterohemorrhagic *E.Coli* (EHEC), *Yersinia enterocolitica* and viral antigen detection for rotaviruses and enteric adenoviruses. The growth media used: Hectoen-enteric, Selenite broth, Sorbitol MacConkey, CIN agar, modified CCDA. Other tests: latex-agglutination, biochemical tests, anti-"O" sera agglutination, Gram stain of culture smear, Vitek identification cards, immunochromatographic tests (viral antigens). Exclusion criteria: positive control stool cultures. Results. There were performed 1998 "routine" stool cultures, 196 "special requests" for *Yersinia*, 108 for EPEC, 24 for EHEC and 1303 rotavirus/adenovirus tests. 185 (9,25%) stool cultures were positive: 89 *Campylobacter* spp., 67 *Salmonella* spp., 13 *Shigella* spp., 9 *Yersinia enterocolitica*, 7 EPEC. 294 (22,5%) viral tests were positive: 80% rotaviruses, 20% adenoviruses. Conclusions. 1.Viral etiology was the most frequent with 2 peaks (May and December). 2.*Campylobacter* spp. was the most frequent bacterial etiology followed by *Salmonella* spp.; 3.Half of *Yersinia* strains were isolated out of "special request" during summer season, suggesting usefulness of adding CIN media in "routine" assay; 4.Authors didn't identified EHEC strain in hemolytic uremic syndrome patients, justifying method revaluation; 5.EPEC strains should be confirmed using molecular tests.

Key words: infectious acute diarrhea, children, stool culture.

56. PROBLEME DE DIAGNOSTIC INTR-UN CAZ DE OBSTRUCTIE GASTRICA LA UN COPIL DE VARSTA SCOLARA

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Introducere: Obstructia evacuarii gastrice implica un spectru larg de afectiuni care perturba pasajul continutului gastroduodenal si evolueaza cu varsaturi persistente non-bilioase.
Metoda: Pacienta CI, de 12 ani, fara antecedente semnificative se interneaza in clinica pentru inapetenta, epigastralgii, varsaturi alimentare, scadere ponderala (5-6Kilograme/luna precedenta) cu tranzit intestinal normal.

Examenul fizic: hipotrofie ponderala (wasting), tegumente palide, varsaturi alimentare intermitent bilioase, dureri epigastrice, fara mase abdominale palpabile.
Investigatiile paraclinice: moderat sindrom biologic inflamator, asociat in dinamica cu dezechilibre hidroelectrolitice si aspect ecografic de steatoza hepatica difuza cu staza gastrica si ingrosare parietala semnificativa.
Esofagogastroduodenoscopia: gastrita flegmonoasa papulo-eroziva cu marcata infiltrare gastrica parietala si deformare antro-pilorica, cu aspect histopatologic si imunohistochimic de gastrita cronica difuza activa non-atrofica, important infiltrat inflamator limfocitar nespecific, Helicobacter pylori negativ, fara proliferari tumorale epiteliale/limfoide.
Evolutia nefavorabila sub antibioterapie parenterala, prokinetice si antisecretoare gastrice a condus la practicarea RMN abdominal care atesta ingrosarea parietala circumferentiala antropilorica extramucoasa cu adenopatii perigastrice intarind suspiciunea etiologica maligna initiala.
Reevaluarea markerilor imunohistochimici din prelevatele bioptice prealabile identifica o proliferare limfoida cu celule B mari/limfom Burkitt CD20+.
Bilantul determinarilor secundare prin computer-tomografie toracica evidentiaza o leziune nodulara la nivelul lobului superior drept incadrata ca tuberculoza pulmonara.
Sub tratament chimioterapic si tuberculostatic evolutia clinica a fost favorabila, cu remiterea partiala a formatiunii tumorale si atenuarea progresiva a obstructiei gastrice.
Concluzii: Cazul prezentat ilustreaza un limfon non-Hodgkin cu localizare gastrica, stadiul III, fara invazia medulara/SNC, localizare rar raportata la copii la care predomina formele ileocecale si care, in mod particular s-a asociat cu tuberculoza pulmonara.
Cuvinte cheie: Obstructie gastrica, limfom non-Hodgkin, copil

DIAGNOSTIC PROBLEMS IN A CASE OF GASTRIC OBSTRUCTION OF A SCHOOL AGE CHILD

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Background: Gastric outlet obstruction encompasses a broad spectrum of conditions that prevent the gastroduodenal contents passage, characterized by persistent nonbilious vomiting.
Methods: A 12 years old girl, without familial/personal history was admitted in our hospital for loss of appetite, epigastric pain, vomiting, weight loss (5-6Kilograms/ previous month), with normal stools.
Physical examination: poor nutritional status (wasting), pale teguments, intermitent bilious vomiting, epigastric pain, without abdominal mass.
Laboratory investigations: moderate inflammatory syndrome associated in dynamics with biological electrolyte imbalances and ultrasound aspect of diffuse liver steatosis with gastric stasis and significant parietal thickening.
Esophagogastroduodenoscopy: papulo-erosive phlegmonous gastritis with marked parietal infiltration and thickened deformed antropyloric region having histological and immunohistochemical features of chronic active non-atrophic gastritis, important nonspecific lymphocytic inflammatory infiltrate without Helicobacter pylori infection and epithelial/lymphoid tumor proliferation.
Parenteral antibiotics, prokinetic and gastric antiacids was initiated with unfavorable evolution, requiring magnetic resonance imaging practice who attest antropyloric circumferential parietal thickening and perigastric lymph node metastases strengthening the suspicion of malignancy.
The reassessment of immunohistochemical markers revealed a large B cell lymphoid proliferation/Burkitt lymphoma CD20+.
Thoracic computed-tomography was performed for the evaluation of secondary determinations and showed a right upper lobe nodular lesion, suggestive for pulmonary tuberculosis.
The clinical evolution was favorable under chemotherapy and tuberculostatic treatment, with partial remission of tumor and progressive attenuation of gastric obstruction.
Conclusions: We presented a case of non-Hodgkin gastric lymphoma grade III without medullary/CNS invasion, rarely reported for children in which the most frequent sites is ileocecal region being particularly associated with pulmonary tuberculosis.
Key words: gastric obstruction, non-Hodgkin lymphoma, children

57. EPIDEMIOLOGIA ANEMIEI IN BOLILE INFLAMATORII INTESTINALE LA COPII: PREVALENTA SI FACTORII ASOCIATI LA DIAGNOSTIC SI LA UN AN

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Obiectiv: Anemia este comuna in bolile inflamatorii intestinale (BII). Studiul analizeaza epidemiologia si factorii asociati anemiei la diagnosticul BII la copii, si la un an de la diagnostic.
Material si metoda: Rezultatele de laborator ale copiilor cu BII, la diagnostic si la un an de la diagnostic, internati intr-o singura clinica universitara, au fost utilizate. Incadrarea anemiei s-a facut conform recomandarilor internationale.
Rezultate: 9 copii (3 fete) cu Boala Crohn (BC) si 12 copii (8 fete) cu colita ulcerativa (CU) au fost luati in studiu. 61,90 % dintre copiii cu BII au fost anemici la diagnosticul bolii si 28,57 % la un an de la diagnostic. 66,66 % dintre copii cu BC si 41,66 % dintre copii cu CU au avut carenta in fier la diagnosticul bolii. La un an, 33,33 % din copii cu BC si 25,0 % din cei cu CU au avut deficienta de fier. Markerii inflamatori au fost pozitivila copii cu anemie la 61,90% dintre acestia.
Conclusion: Prevalenta anemiei la pacientii cu BII la diagnostic este mare. O mare parte din acestia se mentin anemici si la un an de la diagnostic. Sunt necesare eforturi suplimentare pentru tratamentul acestor pacienti cu anemie associate BII.
Cuvinte cheie: anemie, boli inflamatorii intestinale, copii

THE EPIDEMIOLOGY OF ANEMIA IN PEDIATRIC INFLAMMATORY BOWEL DISEASE: PREVALENCE AND ASSOCIATED FACTORS AT DIAGNOSTIC AND FOLLOW-UP ONE YEAR

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Background and aim: Anaemia is common in children with inflammatory bowel diseases (IBD). The study explored the epidemiology and associated factors of anaemia at diagnosis and after 1 year.
Methods: Laboratory results from diagnosis, and at follow up one year were recorded retrospectively in children with IBD recruited from a single tertiary centre. The WHO definition of anaemia was used.
Results: 9 children (3 girls) with Crohn disease (CD) and 12 (8 girls) with ulcerative colitis (UC) were studied. 61,90 % of children with IBD were anaemic at diagnosis, 28,57 % at follow up one year later. 66,66 % of children with CD and 41,66 % with UC were iron deficient at diagnosis. At follow up 33,33 % with CD and 25,0 % of children with UC were iron deficiency. Abnormal inflammatory markers were more common in IBD patients with anaemia (61,90%).
Conclusion: The overall prevalence of anaemia in patients with IBD at the time of diagnosis was high. A large proportion was still anaemic after 1 year. More efforts are needed to treat patients with anaemia.
Key words: anemia, inflammatory bowel disease, children

58. IMPLICAREA INFECTIEI CU HELICOBACTER PYLORI IN BOALA DE REFLUX GASTROESOFAGIAN LA COPII

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Obiectiv: Unele studii sugerează ca infecția cu *Helicobacter pylori (H. pylori)* ar fi un factor de protecție pentru refluxul gastroesofagian. Scopul acestui studiu este de a explora acest fapt.
Material și metode: Un grup de 72 de copii, internați în clinica V Gastroenterologie Pediatrică din Spitalul Clinic de Urgente pentru Copii "Sf. Maria", Iași, diagnosticați cu reflux gastroesofagian prin pH-metrie pe 24h (rezultatele au fost interpretate folosind scorul Boix Ochoa), au beneficiat de endoscopie superioară cu biopsie gastrică.
Rezultate: 19 copii (26.39%) au avut infecție cu *H. pylori*, în timp ce 53 (73.61%) nu au avut. Din 47 de copii cu esofagita I, 16 (34.04%) au avut *H. pylori* prezent, în timp ce, din cei 25 de copii cu esofagita II, doar 3 (12%) au avut *H. pylori*, cu semnificație statistică ($\chi 2 = 54.69$, p << CI 0,05, 95%). Referitor la valoarea scorul Boix Ochoa, se pare că prezența *H. pylori* determină scoruri mai mici (F = 8,13, p = 0,0015, 95% CI).
Concluzii: Prezența *H. pylori* nu a fost un factor important în refluxul gastroesofagian. Pe de altă parte, relația cu esofagita pare a fi invers proporțională. Faptul că prezența *H. pylori* este statistic mai mare în esofagita grad I ar putea confirma ipoteza că acesta ar putea încetini dezvoltarea esofagitei.
Cuvinte cheie: reflux gastroesofagian, *Helicobacter pylori*, pH-metrie, copii

THE IMPLICATION OF HELICOBACTER PYLORI IN GASTROESOPHAGEAL REFLUX DISEASE IN CHILDREN

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Background and aims: Some studies suggests that *Helicobacter pylori* infection would be a protective factor for the gastroesophageal reflux. The aim of this study is to explore this fact.
Methods: A group of 72 children, admitted in V-th Pediatric Gastroenterology Clinic, "Saint Mary"Children Emergency Hospital, Iasi, diagnosed with gastroesophageal reflux by 24 hour continuous esophageal pH monitoring (results were interpreted using the Boix Ochoa score), underwent upper endoscopy with gastric biopsy.

Results: 19 children (26.39%) had *Helicobacter pylori* infection, while 53 (73.61%) did not. Out of 47 children with esophagitis I, 16 (34.04%) had *H. pylori* infection, while out of the 25 children with esophagitis II, only 3 (12%) had *H. pylori* infection, with statistical signifiance (χ^2=54.69, p<<0.05, 95%CI). Regarding the value of the Boix Ochoa score, it appears that the presence of the *Helicobacter pylori* determines lower pHmetry scores (F=8.13, p=0.0015, 95%CI).
Conclusions: The presence of the *Helicobacter pylori* was not an important factor in the gastroesophageal reflux. On the other hand the relation with esophagitis appears to be inverse ratio. The fact that the *Helicobacter pylori* presence is statistically greater in the I-st degree esophagitis could confirm the hypothesis that the bacteria would slow down the development of the esophagitis.
Keywords: gastroesophageal reflux, *Helicobacter pylori*, 24 hour pH-metry, children

59. ASPECTE EVOLUTIVE INTR-UN CAZ DE ACRODERMATITA ENTEROPATICA

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Acrodermatita enteropatica este o boala genetica rara, cu transmitere autosomal recesiva, caracterizata prin dermatita periorificiala, alopecie si diaree. Ea corespunde unui deficit de zinc determinat de un defect de absorbtie intestinala a acestui oligoelement (in particular la nivel duodenal si jejunal). Acesta este datorat unei anomalii cantitative, functionale sau structurale a transportorului intestinal de zinc, Zinc - Binding Ligand Protein 4 (ZIP), proteina produsa de pancreas si codata de gena SLC 39A4 de pe cromozomul 8q24 , la nivelul careia au fost puse in evidenta mai multe mutatii. Prezentam cazul unui baiat in virsta de 1 an şi 2 luni, care s-a internat pentru leziuni eritemato- scuamo- pustuloase , localizate initial inghinal, perianal si pe coapse si leziuni eritematoase foarte discrete la nivelul fetei. Prezenta a numerose suprainfectii cutanate bacteriene cu Stafilococ auriu (inclusiv abcese care au necesitat incizie chirurgicala), au modificat simptomatologia ducind la intirzierea diagnosticului. Leziunile histologice cutanate au fost nespecifice, constind în edem celular, parakeratoza si reducerea stratului granulos. Ulterior s-a conturat un tablou clinic sugestiv pentru aceasta boala care a fost confirmata prin valorile scazute ale zincului plasmatic. S-a initiat terapie cu zinc in doza de 3 mg/ kg/zi de zinc elemental, cu evolutie rapid favorabila, ameliorarea leziunilor cutanate fiind evidenta dupa 2 -3 zile de la inceperea tratamentului. La doua luni de la inceperea tratamentului, leziunile cutanate erau remise complet.
Cuvinte cheie : deficit de zinc, acrodermatita enteropatica.

EVOLUTIONARY ASPECTS IN A CASE OF ACRODERMATITIS ENTEROPATHICA

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Acrodermatitis enteropathica is a rare genetic autosomal recessive transmitted disease, characterized by periorificial dermatitis, alopecia and diarrhea. The disease is the result of a zinc deficiency caused by an intestinal absorption disorder of this micro-nutrient (particularly at the duodenal and jejunal level). This is due to a quantitative, structural or functional abnormality
of the intestinal zinc transporter, Zinc - Ligand Binding Protein 4 (ZIP). This carrier protein is produced by the pancreas and is encoded by SLC gene on chromosome 8q24 39A4; the disease is caused by the mutations in this gene. We present the case of a boy aged 1 year and 2 months,
admitted for erythematous, scaly and pustular lesions, initially located in the inguinal and perianal regions and on the thighs, and very few erythematous lesions on the face. Due to the numerous bacterial skin superinfections with Staphylococcus aureus (including abscesses that required surgical incision), the symptoms modified, leading to a delayed establishment of the diagnosis. The skin histological lesions were nonspecific, consisting in cell swelling, parakeratosis and the reduction of the granular layer. Later, the clinical picture became suggestive for this disease, having been confirmed by low plasma zinc values. Zinc therapy was initiated at a dose of 3 mg/ kg/day of elemental zinc, leading to a rapid favorable evolution: the improvement of the skin lesions few days after the treatment has been started. Two months later, the skin lesions completely disappeared.
Keywords: zinc deficiency, acrodermatitis enteropathica

60. BOALA CELIACA – MANIFESTARI CLINICE SI DIFICULTATI DE DIAGNOSTIC

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Tabloul clinic in boala celiaca este polimorf si multisistemic, de la tulburari digestive pana la stari emotionale schimbatoare.
Va prezentam o serie de cazuri diagnosticate cu boala celiaca in Clinica de Pediatrie – Medicover Bucuresti, in ultimii 5 ani.Manifestarile clasice de boala celiaca au fost uneori discrete, intermitente, instalate de putin timp, uneori sugerate de patologia heredocolaterala (alta decat boala celiaca). Intr-un caz de malabsorbtie au fost luate in considerare particularitatile

imunologice ale copilului sub 2 ani si a fost necesara testarea repetata pentru aceeasi boala, concomitent cu extinderea paletei de investigatii.
Desi testarea/retestarea, pentru aceasta afectiune invalidanta pe termen lung, presupune costuri ridicate, aceasta este necesara la copilul cu suferinta digestiva, inclusiv in cazurile cu tablou clinic mai putin clar.

COELIAC DISEASE-SYMPTOMS AND DIAGNOSTIC CHALLENGE

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Clinical manifestation on coeliac disease are polymorph, ranging from digestive sings to change of mood and emotional distress.We present a series of cases diagnosed with coeliac disease in Pediatric Clinique – Medicover Bucharest, in the last 5 years.
Classic symptoms of the disease were sometimes discreet, random, short timp onset, or suggested by heredocolateral pathology (other than coeliac disease). On one case of malabsorbtion were considered immunologic particularities of under 2 years old child and was necessary retesting for the same disease, in the same time with extension of investigation.
On long term this desease is invalidating one, and even there are important costs for testing/retesting, it should be done for children with digestive suffering, although many times symtoms are not classic.

61. ASOCIERE NEOBISNUITA INTRE POLIPI GASTRICI HIPERPLAZICI SI SINDROM KABUKI LA UN ADOLESCENT

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Introducere: Polipii gastrici, patologie rara la copii, sunt formatiuni tumorale benigne care pot fi identificate imagistic sau endoscopic. Frecventa diagnosticarii in timpul endoscopiei digestive superioare este de 3-5% .
Prezentarea cazului: Un adolescent de sex masculin, de 15 ani se interneaza pentru epigastralgii si varsaturi incoercibile. Din istoric retinem o patologie multipla: sindrom Kabuki, epifizioliza aseptica de cap femural operata osteodistrofie renala severa boala cronica renala terminala, aflat in program de dializa peritoneala. Biologic: anemie normocroma normocitara, sindrom de retentie azotata sever. S-a efectuat endoscopie digestiva superioara care vizualizeaza la nivelul stomacului, prepiloric un pliu gigant de mucoasa cu 2 formatiuni polipoide de 1-1,5 cm, una dezvoltata in portiunea gastrica a pliului cealalta protuzionand intermitent prin pilor in duoden; fara alte aspecte semnificative. La examenul microscopic al biopsiei de mucoasa gastrica nu s-a evidentiat *Helicobacter pylori*. Examenul anatomo-patologic confirma diagnosticul de poplip gastric hiperplazic ascociat cu gastrita antrala cronica usoara, activa.
Concluzii: Polipii hiperplazici se pot asocia cu atrofia mucoasei gastrice cauzata de infectia cu *H. pylori* sau gastrite autoimune. In ultimii ani s-a observat o crestere a cazurilor de polipi asociati cu o mucoasa gastrica normala sau reactiva fara vreo evidenta a infectiei cu *H. pylori*.(2) In cazul nostru particularitatea este reprezentata de aparitia polipilor la un adolescent cu patologie multipla, complexa, asociata. Conduita terapeutica este dificil de stabilit datorita limitelor endoscopiei interventionale; avand in vedere dimensiunea polipilor, in cazul de fata modalitatea optima de tratament ramane cel chirurgical.
Cuvinte cheie: polipi gastrici, copil, endoscopie digestiva superioara

UNUSUAL ASSOCIATION BETWEEN HYPERPLASTIC GASTRIC POLYPS AND KABUKI SYNDROME AT A TEENAGER

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Introduction: Gastric polyps, rare pathology in children, are benign tumors witch can be identified imagistic or endoscopic. The frequency of diagnostic during upper endoscopy is 3-5%.
Case report: A teenager, aged 15 years is admitted for epigastric pain and vomiting. We withhold from his clinical history a wide pathology: Kabuki syndrome, slipped capital femoral epiphysis, severe renal osteodystrophy, Chronic Kidney Disease endstage, in peritoneal dialysis program. Laboratory data: anemia normocytic normochromic, high level of serum creatinine and urea. An endoscopy was performed witch view at the stomach level, before pylorus a giant mucosa crease with 2 polyps with 1-1,5 cm size, one in the gastric portion of the crease the other one passing intermittently from pylorus to duodenum; no other significant aspects. On microscopic examination of gastric mucosa biopsy revealed no *Helicobacter pylori*. Histopathological examination confirms the diagnosis of hyperplastic gastric polyp associated with mild active chronic gastritis.
Conclusion: Hyperplastic polyps may be associated with atrophy of the gastric mucosa caused by *H. pylori* infection or autoimmune gastritis. In recent years we have seen an increase in cases of polyps associated with a normal gastric mucosa or

reactivate without any evidence of infection with *H. pylori*. In our case the particularity is represented by the appearance of polyps at a teenager with multiple and complex pathology associated. Course of treatment is difficult to determine due to the limitations of interventional endoscopy. Given the size of polyps, in this case the best treatment remains surgical.
Key words: gastric polyps, children, endoscopy

62. ULCER GASTRIC TRIPLU, H PYLORI NEGATIV LA UN COPIL DE VARSTA MICA.

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Introducere: Ulcerul peptic este o patologie rar intalnita la copil, si reprezinta o perturbare a integritatii mucoasei stomacului sau a duodenului, cauzata de inflamatie locala, ceea ce duce la intreruperea continuitatii mucoasei digestive. Cauzele majore sunt consumul de AINS, secundar traumatismelor majore sau bolilor sistemice, iar copiii cu parinti fumatori sau care au ulcer peptic, au risc crescut de a dezvolta aceasta patologie.
Prezentarea cazului: Copil de sex masculin in varsta de 8 ani se adreseaza clinicii de gastroenterologie pentru hematemeza si melena. Pacientul nu are istoric de consum de AINS, iar din antecedentele heredocolaterale amintim: parintii cu infectie H Pylori si bunicul patern cu ulcer gastric perforat, operat.
Examenul clinic a relevat obezitate de gradul I; biologic: fara modificari. Grupa sangvina B (III). La endoscopia digestiva superioara se constata: congestie difuza la nivelul 1/3 inferioare a esofagului, cardia competenta. Stomac cu pliuri regulate cu o cantitate minima de suc gastric amestecat cu bila. La nivel prepiloric se vizualizeaza ulcer de forma liniara, cu dimensiuni de aproximativ 2 cm, cu baza edematiata, acoperita cu depozit fibrinos. S-au constatat alte doua ulcere mici, punctiforme, de aproximativ 0,5 cm in diametru, la 2 cm distanta de acesta. Rezultatul biopsiei gastrice: H Pylori negativ.
Examenul anatomopatologic pune diagnosticul de gastrita cronica usoara.
Concluzie: Ulcerul peptic poate duce la complicatii severe, cum ar fi hemoragia digestiva masiva sau perforatia. Particularitatea acestui caz consta in aparitia ulcerului gastric triplu la un copil de 8 ani, cu antecedente personale nesemnificative.
Cuvinte cheie: ulcer gastric multiplu, copil, endoscopie digestiva superioara

TRIPLE GASTRIC ULCER,WITHOUT H PYLORI INFECTION TO AN INFANT

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Introduction: Peptic ulcer disease is a rare pathology in children, it is a disruption of the integrity of the stomach or the duodenum lining. Caused by local inflammation which leads to digestive mucosa continuity interruption. It is caused by the consumption of NSAIDs, major trauma or secondary to systemic illness, and also children who have peptic ulcer as a result of smoking parents have an increased risk of developing this pathology.
Case Presentation: Eight years old boy addresses gastroenterology clinic for hematemesis and melena. The patient has no history of consumption of NSAIDs. The family history include: H pylori infection parents and perforated gastric ulcer paternal grandfather operated. Clinical examination revealed grade I obesity and normal biological tests.Blood group B (III). The upper digestive endoscopy finds: congestion spread to the lower third of the esophagus, cardia competence. Stomach regular folds, with a minimal amount of gastric juice mixed with bile. At the prepiloric area, we visualize an linear shape ulcer, with dimensions of about 2 cm, with an edematous base, covered with fibrinous deposit. They found two other small ulcers, point, about 0.5 cm in diameter, 2 feet away from it. The gastric biopsy is H pylori negative. Pathological examination diagnosis the mild chronic gastritis.Conclusion: Peptic ulcers can lead to severe complications such as gastrointestinal bleeding or massive perforation. The particuliarity of this case lies in the triple stomach ulcers in a 8 years old child, with an insignificant pathological history.
Keywords: triple gastric ulcer, children, endoscopy

63. EFICACITATEA SI TOLERABILITATEA WORMEX IN TRATAMENTUL PARAZITOZELOR INTESTINALE LA COPIL

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Introducere
Parazitozele intestinale reprezinta o categorie de afectiuni frecvent intalnite la copii, acestea fiind adeseori greu de diagnosticat si tratat datorita tabloului clinic nespecific.

Obiective
In acest studiu ne-am propus sa analizam eficacitatea si tolerabilitatea Wormex administrat copiilor cu diverse parazitoze intestinale.
Material si metoda
Am efectuat un studiu clinic, asupra cazurilor pediatrice diagnosticate cu diverse parazitoze intestinale, tratate cu Wormex. La cazurile studiate am monitorizat tolerabilitatea si eficacitatea produsului antiparazitar precum si reactiile adverse survenite. De asemenea am urmarit tabloul clinic precum si varsta, sexul si mediul de provenienta al copiilor. Diagnosticul parazitozelor intestinale a fost stabilit prin examen coproparazitologic. Toate cazurile au primit tratament cu Wormex 30 de zile.
Rezultate
Am luat in studiu 48 copii cu diverse parazitoze intestinale: giardiaza, ascaridiaza si teniaza. Parazitozele intestinale studiate au predominat la sexul masculin, la grupa de varsta 4 – 7 ani, din mediul urban. Simptomatologia prezentata de copii luati in studiu a inclus: dureri abdominale, greturi, varsaturi, inapetenta, cefalee, prurit anal, tulburari ale tranzitului intestinal. Produsul antiparazitar administrat a fost foarte bine tolerat de copiii monitorizati. Reactiile adverse au fost minore, reversibile si nu au necesitat intreruperea tratamentului. Dupa administrarea tratamentului simptomatologia de debut a disparut la majoritatea cazurilor iar examenul coproparazitologic s-a negativat.
Cocluzii
Wormex este un produs antiparazitar foarte bine tolerat de copii, eficient pe principalele parazitoze intestinale pediatrice si poate constitui o alternativa la terapia clasica deoarece are rectii adverse mai putine.

EFFICACY AND TOLERABILITY OF WORMEX IN TREATMENT OF CHILDHOOD INTESTINAL PARASITOSIS

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Introduction
Intestinal parasitosis consists of an array of illnesses frequently found in children whom can be difficult to diagnose and treat because of their unspecific clinical presentation.
Objective
In this study we aimed to analyze the efficacy and tolerability Wormex given to children with various intestinal parasites.
Material and method
We have carried out a clinical study on pediatric intestinal parasitosis cases treated with Wormex. In studied cases we have monitored tolerability and efficiency of the drug as well as possible adverse effects. Clinical presentation, age, sex and background were also monitored. Diagnosis was established through fresh stool examination. All cases received treatment with Wormex for 30 days.
Results
The study comprised of 48 children with various intestinal parasitosis: giardiasis, ascaridosis and teniasis. The most affected were children aged 4-7 years, males and children from urban areas. Symptoms included: abdominal pain, nausea, vomiting, and lack of appetite, headache, anal pruritus, and altered intestinal transit. The anti-parasitic drug was well tolerated in all monitored children. Adverse effects were mild, reversible and did not necessitate interruption of treatment. After treatment, in most cases symptoms disappeared and stool examination was normal.
ConclusionsWormex is an anti-parasitic drug well tolerated in children, efficient in main pediatric parasitosis and can constitute an alternative to classical treatment due to its fewer adverse effects.
Key words: Wormex, child, efficacy, tolerability

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64. DIAGNOSTICUL ”BOALA CELIACA” IN PRACTICA CURENTA PEDIATRICA EXPERIENTA IOMC.

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Introducere
Boala celiaca (BC) este o afectiune sistemica autoimuna produsa prin sensibilizare la gluten la indivizii susceptibili genetic. La copiii netratati boala afecteaza cresterea si dezvoltarea si are potential malign. Pentru sustinerea diagnosticului de certitudine ESPGHAN a elaborat recomandari ferme.
Obiectiv
Ne-am propus evaluarea criteriilor de incadrare in diagnosticul de BC la copiii indrumati catre IOMC pentru monitorizare.
Metode

Un numar de 36 de copii incadrati in diagnosticul BC anterior prezentarii la IOMC au fost evaluati in raport cu recomandarile ESPGHAN din momentul diagnosticului. Criteriile clinice, serologice, histologice si genetice care au condus la diagnostic au fost notate pentru fiecare copil utilizand documente medicale.

Rezultate

Mediana varstelor la diagnosticul BC a fost de 3.6 ani. Dintre acestia, 20 de copii au fost fete. Toti copiii au prezentat criterii clinice la momentul diagnosticului. Pentru 11/36 dintre copii (30.5%) diagnosticul BC a fost stabilit exclusiv prin examen histologic al mucoasei intestinale: o singura biopsie intestinala (BI) la momentul diagnosticului. Pentru 10/36 dintre copii (27.7%) diagnosticul a fost stabilit doar in baza criteriilor clinice si serologice (autoanticorpi specifici) fara a se recurge la BI. La 15/21 copii (71.4%) diagnosticul a fost infirmat ulterior in IOMC prin absenta genelor de risc (HLADQ2/DQ8). Pentru 15/36 dintre copii (41.6%) diagnosticul a fost documentat parcurgand algoritmul de diagnostic recomandat de ESPGHAN.

Concluzii

Diagnosticul BC este in multe cazuri insuficient documentat. Rezultatele noastre subliniaza necesitatea programelor educationale pentru asistenta primara si de specialitate.

Cuvinte cheie: boala celiaca, diagnostic, asistenta medicala

”CELIAC DISEASE” DIAGNOSIS IN PEDIATRIC CLINICAL PRACTICE. IOMC EXPERIENCE.

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2. "Carol Davila" University of Medicine and Pharmacy "

Introduction

Celiac disease (CD) is a autoimmune systemic disease induced by gluten sensitivity in genetically susceptible individuals. Untreated, the disease results in impaired growth and development and potentially leads to malignancies. Currently, pediatric CD diagnosis is supported by ESPGHAN recommendations.

Aim

Our aim was to evaluate the criteria used for pediatric CD diagnosis in current clinical practice.

Methods

Altogether 36 children with previously established CD diagnosis presented at our center for follow-up. The criteria used to diagnose this children were collected from their medical charts and evaluated against ESPGHAN diagnoses guidelines.

Results

Median age at CD diagnosis was 3.6 years and 20 were girls. All children presented symptoms at diagnosis. CD diagnosis was based exclusively on histological examination of duodenal biopsy sample for 11/36 children (30.5%). Another 10/36 children (27.7%) were diagnosed based on clinical symptoms together with celiac-type autoantibody positivity. Consecutively altogether 15 out of this 21 children had prove to be HLA DQ2/DQ8 negatives individuals. The remaining 15/36 children (41.6%) have been diagnosed according ESPGHAN recommended guidelines.

Conclusions

CD diagnosis is still in many cases insufficiently documented. Our results emphasize the need of educational programs for primary and secondary health care system.

Key words: celiac disease, diagnosis criteria, health care system.

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65. ERORI DE DIAGNOSTIC INTR-UN CAZ DE RECTORAGIE LA COPIL

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Introducere:Afectiunile gastrointestinale eozinofilice reprezinta un spectru de boli inflamatorii gastrointestinale caracterizate prin prezenta infiltratelor eozinofilice in peretelui tubului digestiv Aceste afectiuni se clasifica in esofagita eozinofilica, gastroenterita eozinofilica si colita eozinofilica, ultima fiind cea mai rara manifestare descrisa.

Obiective:Lucrarea prezinta un caz de colita eozinofilica la un copil de 3 ani,interpretata la debut ca boala Crohn datorita manifestarilor clinice comune si aspectului endoscopic similar, alaturi de un examen histopatolgic initial neconcludent.

Prezentarea cazului:S.G., sex M,din mediul rural, fara antecedente personale patologice semnificative, la varsta de 3 ani a prezentat un episod de boala diareica acuta cu sange in afebrilitate.Investigatiile efectuate la spitalul teritorial au infirmat etiologia infectioasa.Colonoscopia initiala si examenul histologic desi neconcludent, au orientat catre boala Crohn si s-a initiat corticoterapia orala.La sevrarea corticoterapiei simptomatologia a reaparut,cazul fiind adresat Clinicii noastre pentru completarea investigatiilor. Examenul clinic la internare a fost normal.Investigatiile au aratat eozinofilie periferica, hiperIgE, IgE specifice crescute impotriva cazeinei/alpha,beta lactoglobulinei, calprotectina fecala crescuta.Colonoscopia a evidentiat leziuni aftoide separate de mucoasa normala si inflamatia valvulei ileo-cecale.Examenul histopatologic a aratat la nivelul corionului infiltrat limfocitar si eozinofilic(~30 eozinofile/camp).S-a stabilit diagnosticul de colita eozinofilica asociata alergiei la proteinele laptelui de vaca si s-a initiat dieta de excludere a lactatelor, alaturi de reluarea corticoterapiei, anti-leucotriene si anti-histaminice, cu evolutie favorabila dupa sevrarea corticoterapiei.

Concluzii:O serie de afectiuni intestinale pediatrice pot avea manifestari clinice digestive similare bolii Crohn: enterocolitele infectioase, alergia la proteinele laptelui de vaca, colita eozinofilica.Calprotectina fecala si colonoscopia nu detin acuratete maxima pentru incadrare, examenul histopatologic fiind definitoriu pentru stabilirea diagnosticului.
Cuvinte cheie: Colita eozinofilica,IgE,boala Crohn.

66. GRESELILE DIN ALIMENTATIA SUGARILOR CU INTOLERANTA LA PROTEINELE LAPTELUI DE VACA

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Introducere. Greselile din alimentatia sugarului sunt notate la toate categoriile sociale. Expresia fenotipica a atopiei depinde de interactiunea dintre factorii genetici si de expunerea la alergeni.
Obiectiv. Identificarea erorilor din dieta sugarilor cu alergie la proteinele laptelui de vaca.
Material si metoda. Designul studiului prospectiv cuprinde 52 sugari cu alergie la proteinele laptelui de vaca, diagnosticati timp de un an in Sectia Pediatrie III a Spitalului Municipal ″Dr. G.Curteanu″ Oradea, urmariti pana la varsta de 18 luni. Criteriile de includere au fost cele anamnestice si clinice, respectiv raspunsul la excluderea proteinei incriminate din dieta; testele imunologice s-au efectuat la toți subiectii.
Rezultate. Varsta medie cand a fost decisa alimentatia artificiala a fost 1,86±0,96 luni, initierea alimentatiei mixte s-a efectuat la 2,15±1,18 luni si intarcarea la 3,88±2,60 luni. Acesti sugari au fost alimentati in majoritate cu lapte praf (72%, p=0,01). Diversificarea a fost inceputa in medie la 4,68±0,86 luni, iar introducerea glutenului s-a efectuat in jurul varstei de 5,39±1,55 luni. Aceste practici in dieta sugarilor au determinat manifestari ale intolerantei alimentare, precum urmeaza: cel mai precoce cele cutanate (4±2,58 luni), apoi digestive (4,88±2,92 luni) si respiratorii (6,15±1,81 luni). Alimentatia naturala s-a corelat pozitiv cu amanarea debutului simptomatologiei (r=0,53; p<0,0001), la fel ca varsta intarcarii (r=0,32; p=0,02). Un sfert dintre acesti copii prezinta deficit ponderal in evolutie (p=0,007).
Concluzie. Greselile din dieta sugarului determina intoleranta alimentara precoce cu dificultati ulterioare de alimentatie si de crestere,în strânsă corelație cu precocitatea instituirii alimentației artificiale.
Cuvinte cheie: alimentatie, greseli, alergie

FEEDING ERRORS IN INFANTS WITH COW’S MILK PROTEIN INTOLERANCE

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Background. Feeding errors are noted in infants of any social level, despite of available information regarding the correct diet. The phenotypic expression of atopic diseases depends on interaction between genetic factors and environmental exposure to allergens.
Objective. The aim is to identify the errors in infants feeding in Bihor county.
Material and method. The prospective study evaluated 52 infants diagnosed with cow’s milk protein allergy within one year interval, in the IIIrd Pediatric Department of the Clinical Municipal Hospital “Dr. G. Curteanu” Oradea. Children were followed-up until 18 months of age. Anamnesis, physical examination and dietetic trial were the inclusion criterion for all cases; immunologic tests were performed in each case.
Results. Mean age for bottle-feeding was 1.86±0.96 months, for mixed feeding was 2.15±1.18 months and for weaning was 3.88±2.60 months. The majority of these infants was formula-fed (72%, p=0.01). Complementary feeding was started at 4.68±0.86 months, while gluten was introduced at 5.39±1.55 months. These feeding habits determined clinical manifestations of food intolerance, as following: skin rash (4±2.58 months), then digestive symptoms (4.88±2.92 months) and respiratory ones (6.15±1.81 months). Breastfeeding was positively correlated with the delay of symptoms onset (r=0.53; p<0.0001), as well as the weaning age (r=0.32; p=0.02). A quarter of these children present poor weight gain in evolution (p=0.007).
Conclusion. The errors in infants’ diet determine early food intolerance with further feeding and growing difficulties in correlation with precocity of bottle-feeding.
Key words: feeding, errors, allergy

67. CAUZA RARA DE HIPERAMILAZEMIE LA COPIL

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Prezentam cazul unei fetite in varsta de 3 ani 10 luni, internata in Spitalul Clinic de Copii din Brasov pentru investigarea inapetentei, constipatiei, varsaturilor ocazionale, debutate de la varsta de 2 ani cu rezolutie intermitenta.

La internare examenul clinic a evidentiat un copil afebril, fara adenopatii superficiale, cu glande salivare nepalpabile, echilibrat cardiovascular si respirator, un abdomen suplu, nedureros spontan sau la palpare, fara aparare abdominala, fara pierderi digestive, tranzit intestinal incetinit, apetit diminuat. Investigatiile de laborator si imagistice efectuate pentru suspiciunea unei afectiuni digestive au fost normale, cu exceptia amilazei serice marcat crescute, cu lipaza serica si amilazuria normale. Interpretarea CTului abdominal a sugerat un posibil diagnostic de pancreatita acuta, nesustinut de ecografiile abdominale seriate.
In evolutie, copilul s-a mentinut cu stare generala satisfacatoare, fara dureri abdominale, cu rezolutia inapetentei si constipatiei sub tratamentul pentru afectarea pancreatica. Amilaza serica s-a mentinut crescuta. S-au efectuat: raportul clearance amilaza/creatinina, macroamilaza serica, izoenzimele amilazei (amilaza salivara crescuta), pe baza carora s-a stabilit diagnosticul de macroamilazemie (MA). S-au exclus alte afectiuni care pot asocia MA.
Discutii: Macroamilazemia este o afectiune benigna, extrem de rara in populatia pediatrica, caracterizata prin prezenta de complexe macromoleculare (amilaza pancreatica/salivara legata de imunoglobulina tip IgG/IgA), greu eliminate pe cale renala datorita greutatii moleculare mari. Peristenta complexelor in ser determina nivele crescute ale amilazei serice.
Amilaza serica crescuta in context de simptomatologie digestiva, in absenta afectarii pancreatice acute, trebuie sa conduca la suspiciunea MA si la copil, in vederea evitarii investigatiilor costisitoare, invazive si a tratamentului inutil.

A RARE CAUSE OF HYPERAMYLASEMIA IN CHILDREN

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We report a case of a 3-year-old girl admitted in our hospital poor appetite, constipation and occasional vomiting with onset since the age of 2 years with incomplete remission.
She had normal body temperature and body weight, HR=100 beats per min, clear breath sounds. There was decreased appetite despite no evidence of palpable lymph nodes or salivary glands, abdominal pain, vomiting. Laboratory and imaging tests were normal except for high serum amylase (905U/L) with normal serum lypase and amylasuria. Abdominal CT-scan was interpreted as suggestive for acute edematous pancreatitis, despite several normal abdominal ultrasound examinations. Under therapy, she felt well, with no abdominal pain, resolution of constipation and improved apetite. As serum amylase value continued to be abnormally high (>700U/L) we performed investigations to rule out other causes of hyperamilasemia. The final diagnosis was macroamylasemia (MA), based on amylase/creatinine ratio, serum macroaymlase identification and high salivary serum amylase. We also excluded other conditions that may be associated with MA.
Discutions: Macroamylasemia is a benign condition, extremely rare among pediatric patients. MA is caused by circulating macromolecular complexes consisting of pancreatic or salivary amylase bound to plasma immunoglobulins (Ig G/Ig A). Their large size impair renal filtration and result in hyperamylasemia. High serum amylase, in absence of pancreatic disease, must lead to suspicion of MA, in order to avoid unnecesary and expensive investigations and treatment.

68. FISTULELE POSTOPERATORII IN TRATAMENTUL DESPICATURII PALATINE

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Introducere: Fistula postoperatorie este o complicatie semnificativa dupa repararea chirurgicala a palatoschizisului.
Scopul acestui studiu retrospectiv este de a determina incidenta fistulelor postoperatorii in tratamentul despicaturii palatine si de a evidentia factorii predictivi ai aparitiei acestor complicatii.
Materiale si metode: Au fost inclusi in studiu 30 pacienti operati in clinicile de chirurgie pediatrica si chirurgie plastica ale Spitalului "Grigore Alexandrescu" Bucuresti intre anii 2005 si 2008. Au fost selectati numai pacientii cu forma nonsindromica de despicatura palatina izolata. S-a efectuat analiza statistica utilizand Microsot Excel 2010 pentru determinarea factorilor ce conduc la cresterea riscului de aparitie a fistulei postoperatorii.
Rezultate: In perioada mentionata au fost operate 14 cazuri cu forma incompleta Veau I si 16 cazuri cu forma completa Veau II, 21 dintre acestea intr-o singura etapa si 9 in doi timpi. Incidenta generala a formari fistulelor palatine a fost de 20%. Au fost evidentiati cativa factori predictivi de fistulizare: tratamentul chirurgical in doua etape cu o rata de fistulizare de 66,66%, varsta mica la a doua etapa a interventiei chirurgicale, sexul feminin si forma completa a despicaturii palatine(Veau II). S-a constatat o incidenta crescuta a cazurilor operate la varsta mare in cazul pacientilor cu domiciliu rural, corespunzatoare unei educatii medicale precare.
Concluzii: Studiul efectuat demonstreaza o incidenta relativ crescuta a fistulelor palatine dupa tratamentul chirurgical al palatoschizisului, in special atunci cand e utilizat procedeul in doua etape.
Cuvinte cheie: despicatura palatina , fistula
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POSTOPERATIVE FISTULA IN CLEFT PALATE SURGERY

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Introduction: Postoperative fistula is a significant complication associated with cleft palate surgery.
The objective of this retrospective study is to establish the incidence of postoperative fistula in cleft palate surgery and to highlight the predictive factors for the occurence of this complication.
Materials and methods: We studied 30 patients reffered to pediatric surgery and plastic surgery departments of "Grigore Alexandrescu" Hospital, Bucharest between 2005 and 2008. There were selected only non-syndromic isolated cleft palate patients. For the statistic analysis we used Microsoft Excel 2010 to establish all contributing factors for an increased risk of postoperative fistula.
Results: During the mentioned time, 14 cases of incomplete form (VeauI) and 16 cases of complete form (VeauII) underwent surgical interventions. Of all these, 21 cases had one-stage palate repair and 9 cases were treated by 2-stages surgery. The general incidence of fistula was 20%. We can highlight several predictive factors for fistula: 2-stage surgery with a fistula rate of 66,66%, a smaller age in the second stage of surgical intervention, female sex and the presence of a complete form (VeauII). We observed an increased number of cases who underwent surgical intervention later in life and came from country sites, meaning a poor medical knowledge.
Conclusions: This study showed a relative increased incidence of palate fistula after cleft surgery, especially when 2-stage surgery is used.
Key words: cleft palate, fistula

69. EVALUAREA EFICACITĂŢII TRATAMENTULUI CU SER FIZIOLOGIC LA SUGAR ŞI COPILUL MIC, CU SINDROM DE DESHIDRATARE ACUTĂ >10 %

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Introducere: rehidratarea hidroelectrolitica parenterala reprezinta cheia tratamentului în diareea acuta severa.Terapia cu ser fiziologic in bolus este cea mai utilizata în pofida raportarii, în cadrul studiilor clinice internationale, a efectelor ei adverse.
Scopul studiului: obiectivul principal este evaluarea eficientei tratamentului cu ser fiziologic in bolus in sindroamele de deshidratare acuta (SDA) >10 % , implementarea noilor protocoale de tratament.
Pacienti şi metodă: studiul efectuat este retrospectiv, pe 74 de copii cu vârsta < 12 luni, diagnosticati cu SDA >10 % in Spitalul Clinic de Copii Braşov, in perioada 2006-2013.
Rezultate şi discuţii: 66% pacienţi sunt de sex masculin, 34% sex feminin, 62% provin din mediul urban, 38% mediul rural şi au media de varstă 4,97 luni. Markerii inflamatori indică etiologia virala iar valorile creatininei si ureei insuficienţa prerenală. In 97% din cazuri etiologia este neprecizata. Sub tratament valorile sodiului, calciului si potasiului se menţin in limite normale, insa valorile clorului sunt crescute. Evaluarea parametrilor ASTRUP ne indica urmatoarele valorile medii înainte de tratament : pH 7.25, HCO3 13.47,Beb -12.79, BEecf -12.1707 iar dupa administrarea de albumina sau plasma sunt de : pH 7.34, HCO3 19.6, Beb -5,4 BEecf -6.2, in timp cu ser fiziologic numai valoarea pH-ului se modifică restul parametrilor ramanand neschimbayi.
Concluzii: datele acestui studiu sugereaza ca administrarea serului fiziologic in bolus creste numarul zilelor de spitalizare.

EVALUATION OF THE EFFICACY OF SALINE TREATMENT FOR INFANTS AND TODDLERS WITH SEVERE DESHYDRATION OVER 10%

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Introduction: intravenous rehydration represents the key for treatment of severe dehydration at infants and toddlers. Bolus of saline is the most used one despite recent literature reports regarding its adverse effects.
Aim of the study: the main objective of our study was to evaluate the efficacy of saline boluses in severe dehydration due to diarrhea as well as to implement new protocols regarding this issue.
Patients and methods: retrospective study, on 74 infants and toddlers that completly fullfilled the criteria of severe dehydration over 10% admitted at the Childrens Clinic Hospital, Brasov, during 2006-2013.
Results and discussions: 66% were male patients, 34% of them are girls and 62% come from urban area, mean age at admittance was 4.97 months. Inflammation markers were in normal rangewhile ureas and creatinines values expressed a

prerenal insufficiency. In 97% of the cases etiology remained unknown. Normal values of ions: Natrium, Pottasium and Calcium while Chlorum was high.
ASTRUP values showed following values before traeatment: pH 7.25, HCO3 13.47,Beb -12.79, BEecf -12.17 while after treatment with albumin or plasma they were: pH 7.34, HCO3 19.6, Beb -5,4 Becf-6.2. In case of saline only pH values will modify while the rest of ASTRUP remains unchanged.
Conclusion: data of this study suggest that administration of physiologic serum increase the number of hospitalization days.

70.TULBURARI DE MOTILITATE GASTRO-INTESTINALA LA COPIL

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Introducere: Relația dintre constipatie si boala de reflux gastroesofagian este neclara. Doar cateva studii au evaluat asocierea la copii și adolescenți. Din acest motiv ne-am propus să analizăm frecventa bolii de reflux gastroesofagian la copiii cu constipatie cronica funcțională.
Material și metoda: Grupul de studiu a constat din 994 de copii diagnosticati cu constipatie, cu varsta cuprinsa intre 1-17 ani, internați în serviciul de Gastroenterologie, Spitalul de Urgenta pentru Copii din Iasi, în perioada 1 ianuarie 2012 - 1 ianuarie 2014. Constipația a fost definita conform criteriilor Roma III. Toți pacienții incluși în studiu au raspuns la un chestionar clinic referitor la simptomatologia legate de boala de reflux, și apoi la toti pacientii simptomatici s-a efectuat tranzit baritat .
Rezultate: Lotul de studiu a reprezentat 9,6% din totalul copiilor spitalizati. În ceea ce privește prevalența constipatiei nu am observat o diferență semnificativă între sexe, dar am găsit o proportie semnificativa de pacienti provenind din mediul urban 68% (676/994), comparativ cu 32% (318/994) din mediul rural. Prevalenta obezitatii a fost de 11,46% (114/994) din populatia de studiu. Conform tranzitului baritat, refluxul patologic a fost identificat la 36,73% (72/196) dintre pacienți. Nu am gasit nici o diferenta semnificativa referitoare la vârstă, sex și durata constipatiei.
Concluzii: Constipatia cronica este o afectiune comuna, care afecteaza copiii de toate varstele. Boala de reflux gastroesofagian ar trebui luata în considerare în tratamentul și monitorizarea pacienților cu constipație funcțională
Cuvinte-cheie: Constipație; Boala de reflux gastroesofagian; Copil

GASTROINTESTINAL MOTILITY DISORDERS IN CHILDHOOD

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Background: The relation between constipation and gastroesophageal reflux is unclear. Only few studies have evaluated the association in pediatric population. We aimed to analyze the frequency of gastroesophageal reflux disease in children with functional constipation.
Materials and methods: The study group consisted of 994 children diagnosed with constipation, aged from 1 to 17 years old, hospitalized in the Gastroenterology Service, "Saint Mary" Emergency Hospital for Children of Iasi, between 1 January 2012 and 1 January 2014. Constipation was defined according to Rome III criteria. All patients included in the study were submitted to a clinical questionnaire about reflux-related symptoms, and then all symptomatic cases underwent a barium swallow.
Results: The study group represented 9.6% of all hospitalized children. Regarding the prevalence of constipation we did not notice a significant difference between the sexes, but we found a significant proportion of patients from urban areas 68% (676/994) versus 32% (318/994) in rural areas. The prevalence of being overweight was found in 11,46% (114/994) of the study population. According to the barium swallow, pathologic acid reflux was determined in 36.73% (72/196) of the patients. No significant difference was found in terms of age, gender and duration of constipation.
Conclusions: Constipation is an extremely common problem that affects children of all ages. Gastroesophageal reflux disease should be considered in the treatment and monitoring of patients with functional constipation
Keywords**:** Constipation; Gastroesophageal reflux disease; Child

71. INFECȚIA CU VIRUSUL EPSTEIN BARR ȘI COLECISTITA ALITIAZICĂ, O ASOCIERE RARĂ LA UN PACIENT PEDIATRIC

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Colecistita acută alitiazică (CAA) asociată cu infecția virusului Epstein Barr (vEB) este o entitate cunoscută, dar stenoza ductului cistic secundară inflamației cauzate de vEB este un caz extrem de rar. Autorii prezintă cazul unui pacient în vârstă de 10 ani de sex masculin, care se prezintă (noiembrie 2012) la spital cu stare generală alterată, anorexie, meteorism abdominal, abdomen dureros fiind internat în Clinica de Chirurgie Pediatrică. Din antecedentele patologice menționăm că a fost diagnosticat în 2011, cu hepatită acută cu vEB. Pe durata spitalizării a acuzat febră, vărsături, dureri severe la nivelul hipocondrului drept iar după consultul chirurgului pediatru și a ecografiei abdominale care descrie CAA cu hidrops vezicular (HV) s-a decis temporizarea intervenției chirurgicale. Ulterior, evoluția clinică a fost favorabilă, dar în decursul celor doi ani copilul a suferit două episoade de exacerbare a colecistitei cronice alitiazice (CCA) și menține HV. Evoluția clinică a fost favorabilă dar persistența HV a necesitat efectuarea unui examen RMN, care a evidențiat HV și îngustarea segmentului terminal al ductului cistic (secundar procesului inflamator). La momentul actual investigațiile biologice au relevat sindrom inflamator, colestază și hepatocitoliză. Ecografia abdominală descrie colecist alitiazic și HV (dimensiuni de 80\48 mm). Tratamentul a fost chirurgical, reprezentat de colecistectomie. Examenul histopatologic evidențiază inflamație cronică a pereților colecistului și stabilește diagnosticul de exacerbare a CCA. Particularitatea cazului este că, după infecția acută cu vEB copilul a dezvoltat o stenoză a ductului cistic. Evoluția clinică și paraclinică în dinamică (după 1, 3, 6 și 12 luni) a fost favorabilă.
Cuvinte cheie: colecistită alitiazică, infecție cu virusul Epstein - Barr, colecist, hidrops, copil

EPSTEIN BARR VIRUS INFECTION AND ACALCULOUS CHOLECYSTITIS, A RARE ASSOCIATION IN A PEDIATRIC PATIENT

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Acute acalculous cholecystitis (AAC) associated with Epstein Barr virus (EBV) infection is a known entity, but the cystic duct stenosis secondary to inflammation caused by EBV is an extremely rare case. The authors present the case of a 10-year-old male patient who comes (November 2012) at hospital with altered general state, anorexia, abdominal meteorism, painful abdomen and he is admitted at Pediatrics Surgery Department. From the case history we reveal that he was diagnosed in 2011 with EBV acute hepatitis. During that hospitalization he presenting fever, vomiting, severe pain of right upper abdominal quadrant and after a clinical examination of pediatric surgeon and abdominal echography revealing AAC with gallbladder hydrops (GH) the surgery was postponed. Subsequently, the outcome was favorable but during those two years he suffered two episodes of exacerbation of chronic acalculous cholecystitis (CAC) and maintaining the GH. Clinical outcome was favorable but the maintaining of GH required performing an MRI examination which revealed GH and narrowing of terminal segment of the cystic duct (secondary inflammatory processes). At this admission the biological investigations revealed cholestasis, hepatocitolysis and inflammatory syndrome. The abdominal-echography reveals acalculous gallbladder and GH (80\48mm in dimensions). The treatment was surgical and consisted in cholecystectomy. The histopathology examination reveals chronic inflammation of the gallbladder wall and sets the diagnosis of exacerbation of CAC. Particularity is that after the EBV acute infection the child has developed a cystic duct stenosis. The clinical and laboratory data in dynamics (after 1, 3, 6 and 12 months) was favorable.
Keywords: acalculous cholecystitis, Epstein - Barr virus infection, gallbladder, hydrops, child.

HEMATOLOGIE - ONCOLOGIE / HAEMATOLOGY - ONCOLOGY

72.TUMORA NEDIFERENTIATA CU METASTAZE MULTIPLE LA SUGAR

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Tumorile intraabdominale maligne la sugar se caracterizeaza prin lipsa, in cele mai multe cazuri, a aspectelor clinice specifice care pot ajuta la diagnosticul diferential precoce, detectarea in stadii avansate afectand supravietuirea. Prezentam cazul unui sugar in varsta de 2 luni, de sex masculin, internat pentru febra, tuse productiva, tranzit intestinal incetinit. Clinic la internare prezenta tegumente palide, cu circulatie colaterala la nivel abdominal, dispnee, tiraj generalizat, raluri crepitante si subcrepitante bilateral, abdomen destins de volum, ficat la 3 cm sub rebord, hidrocel drept. Biologic: neutrofilie, limfopenie,

anemie intrainfectioasa si discreta citoliza hepatica. Radiografia toracica a evidentiat multiple opacitati nodulare, diseminate in ambele arii pulmonare, sinus costodiafragmatic drept opac. S-a stabilit diagnosticul de etapa de bronhopneumonie si s-a instituit terapie antibiotica si simptomatica timp de 10 zile, cu evolutie insa nefavorabila: aparitia icterului, a hepatosplenomegaliei, ascitei, leucopeniei, trombocitopeniei si accentuarea anemiei, medulograma fiind normala. Ecografia abdominala a obiectivat imagini hipodense la nivel splenic si hepatic. In acest context s-a decis efectuarea unui CT toraco-abdominal, ce a evidentiat marirea difuza de volum a muschilor psoas si paravertebrali stangi, printr-o formatiune imprecis delimitata, ce invada canalul medular, multiple leziuni nodulare in parenchimul pulmonar, hepatic si splenic. Decesul a survenit inainte de initierea terapiei specifice. Necropsia a confirmat prezenta tumorii descrise, cu invazia canalului medular, si leziuni metastatice in pulmon, ficat, splina, testicol. Anatomo-patologic s-a evidentiat o tumora cu celule “mici albastre” in bisac, nediferentiate, pentru precizarea originii celulei tumorale fiind necesare studii imunohistochimice. In prezenta metastazelor multiple si a caracterului nediferentiat, tumorile maligne ale sugarului au prognostic nefavorabil.

UNDIFFERENTIATED TUMOR WITH MULTIPLE METASTASES IN INFANT

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Abdominal malignant tumors in infants, in most cases, are characterized by a lack of specific clinical signs that may help in the early differential diagnosis, detection in advanced stages affecting survival. We present the case of an infant aged 2 months, male, hospitalized for fever, productive cough, slowed intestinal transit. At admission, the child had paleness, collateral circulation in the abdomen, shortness of breath, bilateral crackles and crepitation, distended abdomen, liver at 3 cm below the lip, right hydrocele. Biological: neutrophilia, lymphopenia, intrainfectious anemia and discrete hepatic cytolysis. Chest radiography showed multiple nodular opacities disseminated in both lung fields, right costodiafragmatic sinus opaque. The diagnosis was of bronchopneumonia and the patient received antibiotic therapy and symptomatic for 10 days, but the evolution was unfavorable: appearance of jaundice, hepatosplenomegaly, ascites, leukopenia, thrombocytopenia and accentuation of anemia, with normal medulogram. Abdominal ultrasound showed hypodense images in the liver and spleen. In this context it was decided to perform an thoracic-abdominal CT, which revealed diffuse enlargement of psoas and left paraspinal muscle through a imprecisely delimited tumor, that invades the spinal canal, and multiple nodular lesions in the lung parenchyma, hepatic, splenic. Death occurred prior to initiating specific therapy. The autopsy confirmed the presence of described tumor, with spinal canal invasion and metastatic lesions in the lung, liver, spleen, testicle. Histopathological review reveal a tumor with "little blue" cells, undifferentiated; for specifying the origin of tumor cell immunohistochemical studies are needed. Malignant tumors with multiple metastases and undifferentiated nature have poor prognosis in infants.

73. TROMBOCITOPENIE CRONICA LA SUGAR - FORMA DE PREZENTARE A SINDROMULUI WISKOTT-ALDRICH

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Scopul si obiectivele studiului. Prezentarea unui caz de trombocitopenie cronica la un sugar diagnosticat relativ precoce cu o afectiune genetica cu transmitere X-linkata extrem de rara (OMIM 301000) si potential fatala - sindromul Wiskott-Aldrich (WAS)
Material si metode. Pacientul M.F., sex masculin, nascut in 28.04.2011, a fost diagnosticat cu WAS la varsta de 7 luni (noiembrie 2011, Prof.Marodi Laszlo, Debrecen); la varsta de un an si 11 luni (21.03.2013) pacientul a primit un transplant autolog de celule stem modificate genetic (Prof.Alessandro Aiuti, Milano) cu evolutie ulterioara favorabila.
Rezultate si discutii. Pacientul a prezentat de la varsta de 6 saptamani trombocitopenie persistenta si episoade recurente de diaree snaguinolenta, dermatita eczematiforma, foliculita, conjunctivita, candidoza bucala; la varsta de 6 luni, pe baza manifestarilor clinice si a modificarilor biologice progresive (aparitia microtrombocitelor, scaderea limfocitelor T CD8+, a celulelor NK si ulterior si a limfocitelor B CD4+, inversarea raportului CD4/CD8, aparitia hipogamaglobulinemiei, a neutropeniei posibil autoimuna) s-a ridicat suspiciunea de WAS; confirmarea diagnostica a fost rapida prin evidentierea absentei proteinei specifice (WASp), ulterior identificandu-se si anomalia genica - deleţie extinsă la nivelul genei WAS (ianuarie 2012); mama a fost identificata ca purtatoare a mutatiei. Pe baza scorului clinc de severitatate (Ochs HD, 2009) boala a fost incadrata ca forma severa, cu indicatie absoluta de transplant de celule stem la varsta mica.
Concluzii. Diagnosticarea relativ precoce a WAS a permis interventia terapeutica rapida cu evolutie favorabila pana in prezent, la 2 ani si 2 luni de la transplant.

Cuvinte cheie: Trombocitopenie, WAS

CHRONIC THROMBOCYTOPENIA IN INFANT - PRESENTING MANIFESTATION OF WISKOTT-ALDRICH SYNDROME

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Scope and objectives. Presentation of a chronic thrombocytopenia case in an infant relatively early diagnosed with an extremely rare X-linked genetic disorder (OMIM 301000) potentially fatal - Wiskott-Aldrich syndrome (WAS).
Material and methods. The male patient M.F., born on 28.04.2011, was diagnosed with WAS at the age of 7 months (November 2011, Prof.Marodi Laszlo, Debrecen); at the age of 6 months (21.03.2013) the patient received an autologous transplant with genetically modified stem cells (Prof.Alessandro Aiuti, Milano), with a good evolution.
Results and discussions. The patient had from the early age of 6 weeks persistant thrombocytopenia, and recurrent episodes of bloody diarrhea, eczematous dermatitis, folliculitis, conjunctivitis, oral Candidiasis; at the age of 6 months, based on the clinical picture and on the progressive biological abnormalities (microthrombocytopenia, reduction of CD8+ T cells, of NK cell, and further of CD4+ B cells, increased CD4/CD8 ratio, hypogammaglobulinemia, neutropenia) the suspicion of WAS was taken into consideration; there was a rapid diagnostic confirmation by the absence of WAS protein (WASp); the gene mutation was further identified - large deletion of the WAS gene (January 2012); the mother was foun to be carrier for the same mutation. Based on the clinical severity score (Ochs HD, 2009), the disease was cosidered as a severe form, with the absolute indication of stem cell transplantation at an early age.
Conclusions. The relatively early diagnosis of WAS permitted the rapid therapeutical intervention with a favourable response until present time, 2 years and two months after transplantation.
Key words: thrombocytopenia, WAS

74. SINDROMUL PEUTZ JEGHERS CAUZA UNEI ANEMII PERSISTENTE

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Peuts Jeghers este un sindrom autosomal dominant , dar sunt descrise si cazuri de novo. Se caracterizeaza prin asocierea polipozei gastro-intestinale, pigmentarea mucoaselor si risc crescut de malignizare. Simptomatologia este adesea cu dureri abdominale recurente, anemii persistente la tratament, ocluzii intestinale.
Material si metode: Pacient in varsta de 6 ani 8 luni se prezinta pentru dureri abdominale si varsaturi asociate cu sindrom anemic sever, refractar la terapia martiala. La examenul clinic se deceleaza macule hipercrome la niveul mucoasei bucale, fara focar de sangerare. Datele praclinice confirma anemia hipocroma, hiposideremica izolata, evidentiaza sangerare digestiva oculta. Examenul colonoscopic pune in evidenta polip rectal cu aspect hisopatologic de polip glandular. Ecografia, examenul cu capsula endoscopica , entero RMN deceleaza polipi la nivelul intestinului subtire, care evidentiaza fenomene de invaginatie intermitenta. Se intervine chirurgical, se practica rezectie segmentara jejun-ileon se deceleaza anse purtatoare de 4 polipi cu dimensiuni intre 1/1 cm si 4/4 cm. Anatomia patologica sustine diagnostciul de Sindrom Peutz – Jeghers.
Conculzie : Avand in vedere complicatiile acetui sindrom , se impune monitorizare anula a hemoglobinei si la interval de 2 ani , endoscopie eso-gastrica si colonoscopie.

PEUTZ JEGHERS SYNDROME – SEVERE ANEMIA'S ETIOLOGY

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Peutz Jeghers is autosomal dominant genetic diseas but there are new de novo cases reported. Thes condition is characterized by the association of gastrointestinal polyposis, mucocutaneous pigmentation and cancer predisposition. It is described by recurrent abdominal pain, persistent anemia unresponsive to any treatment and bowel obstruction.
Methods: 6 year and 8 month patient presenting for abdominal pain, vomiting associated with sever anemia unresponsive iron therapy. Clinical exam reveals hiperpigmented macules around the oral mucosa without any source of bleeding. Paraclinical investigations confirm hypocrom, hyposideremic anemia and schous occult gastrointestinal bleeding. The colonoscopy reveals a rectal polyp with a glandular histopathological aspect . The ultrasound , capsule endoscopy, and entero IMR examination shous small- bowel polyps causing intermitent intussusception. During surgical procedure reveals 4 polyps with dimension between 1/1 – 4/4 cm. The hystophatologycal exam sustain the Peutz Jeghers Syndrome diagnosis.
Conclusion: Considering complications of this syndrome, it is mandatory an annual checking of the hemoglobin values and every 2 years endoscopy and colonoscopy examination.

75.CONSIDERATII DESPRE DOI PACIENTI CU MEDULOBLASTOM

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Meduloblastomul, cea mai frecventa tumora cerebrala la copil, este deseori localizata la nivelul cerebelului (in fosa posterioara). Ca factori de risc au fost citati: radiatiile ionizante, virusuri ca Epstein-Barr, prezenta unor boli genetice ca neurofibromatoza.

Autorii prezinta doua paciente: MDM- copil mic de un an si trei luni si VF, in varsta de trei ani si opt luni in momentul diagnosticarii. Amandoua au avut astazie, tulburari de echilibru, inapetenta, au emis varsaturi de tip central; VF a prezentat si o criza de convulsii in afebrilitate. Cele doua paciente proveneau din genitori tineri, sanatosi, doar la MDM notandu-se in A H-C o strabunica cu neoplasm genital.

Tomografia craniana a relevat o tumora de fosa posterioara la MDM, respectiv o tumora de vermis la pacienta VF.

Examenul anatomopatologic din biopsia prelevata preoperator la M.D.M, respectiv recoltata intraoperator la VF , impreuna cu imunohistochimia, au stabilit diagnosticul de meduloblastom cerebral nodular/desmoplazic in ambele cazuri.

La pacienta V.F s-a intervenit neurochirurgical ca pentru o tumora benigna , insa era meduloblastom cu celule slab diferentiate, in stadiul IV . S-au depistat recidive cerebrale, metastaze meningiene; chimioterapia n-a fost tolerata, pacienta decedand la patru luni de la operatie.

La MDM s-a introdus precoce chimioterapia, ablatia chirurgicala facandu-se dupa punctia-biopsie. Pacienta are un an de la diagnosticare, este euponderala, a suportat bine curele de chimioterapie si are investigatii normale .

Meduloblastomul este o tumora maligna agresiva, cu evolutie rapida: in ciuda tratamentului corect, doar 47% dintre pacienti supravietuiesc peste cinci ani.

CONSIDERATIONS ON TWO PATIENTS WITH MEDULLOBLASTOMA

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Medulloblastoma, the most common brain tumor in children, is often located in the cerebellum (the posterior fossa). As risk factors were cited: ionizing radiation, Epstein-Barr virus, the presence of genetic diseases like neurofibromatosis.

The authors present two patients: MDM- todler, a year and three months age, and VF, aged three years and eight months at diagnosis. Both had inability to stand up, postural balance disorders, loss of appetite, vomiting issued by central type; VF has presented a crisis of seizures without fever. The two patients came from genitors young, healthy, noting only MDM in A HC a grandmother with genital cancer.

Cranial tomography revealed a posterior fossa tumor in MDM and a tumor of vermis in VF patient.

Pathological examination of the biopsy sample taken preoperative from the MDM or intraoperative harvested for VF, along with immunohistochemistry, were diagnosed cerebral nodular medulloblastoma in both cases.

The neurosurgical patient VF intervened as a benign tumor, but it was poorly differentiated cell medulloblastoma in 4-th stage. Recurrences were detected in brain and meningeal metastases; chemotherapy was not tolerated, patients dying four months after surgery.

In MDM was introduced early chemotherapy, surgical ablation being after done biopsy. The patient has a year of diagnosis, is eutrophic, well supported courses of chemotherapy and has normal investigations.

Medulloblastoma is an aggressive malignant tumor with rapid evolution: in spite of correct treatment, only 47% of patients survive more than five years.

Key-words: Medulloblastoma, children, brain

76. PROBLEME DE DIAGNOSTIC SI TRATAMENT INTR-UN CAZ DE LEUCOCITOZA PERSISTENTA

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Obiective

Prezentarea principalelor probleme de diagnostic in cazul unui copil copil mic, cu numeroase internari in antecedente ce aveau in comun o leucocitoza persistenta.

Pacient, metoda, rezultate

Pacientul I.G.A in varsta de 21 luni se interneaza pentru simptomatologie respiratorie in context febril. Investigatiile de laborator indica o leucocitoza marcata si o trombocitoza importanta. Aceasta modificare a hemoleucogramei se mentine in ciuda tratamentului instituit. Se exclud cauzele cele mai frecvente de leucocitoza pentru aceasta categorie de varsta: infectii, inflamatii, reactii alergice. Consultul hematologic exclude o hemopatie maligna.

Din istoricul pacientului retinem ca la determinari repetate ale hemoleucogramei s-au inregistrat aceleasi modificari: leucocitoza persistenta asociata cu o trombocitoza importanta. De fiecare data frotiul de sange periferic indica poikilocitoza cu ovalocite, schizocite, eritrocite in picatura, anizocitoza cu microcitoza.

Ecografii abdominale repetate la intervale de timp diferite si de catre examinatori diferiti nu vizualizeaza splina. Acest fapt, asociat cu modificarile hemoleucogramei si ale hematiilor pe frotiul de sange periferic, ridica suspiciunea unei asplenii congenitale.
Metodele de cuantificare ale functiilor splenice au la baza parametrii hematologici, imunologici si scintigrafici.
Ca metode imagistice s-au efectuat ecografia abdominala si TC cu substanta de contrast.
Concluzii Ca urmare a investigatiilor efectuate s-a stabilit diagnosticul de asplenie congenitala izolata.Managementul cazului in conditiile suspectarii unei infectii este deosebit de important deoarece acesta este supus riscului infectiilor fulminante si necesita profilaxie datorita riscului de infectie cu germeni capsulati.
Cuvinte cheie:leucocitoza, trombocitoza, asplenie congenitala

DIAGNOSIS PROBLEMS AND TREATMENT IN A CASE OF PERSISTENT LEUKOCYTOSIS

Dr Roxana Florescu, Dr Adela Radulescu, Dr Ghita Anca, Dr Florin Brezan, Prof.Dr. Matei Dumitru

Objectives
Presenting the main diagnosis problems in the case of an infant child, known with a history of numerous hospitalisations which had in common persistent leukocytosis.
Patient, methods, results
A 21 months old patient I.G.A, is hospitalised with febrile respiratory symptoms.
The laboratory investigations display a noticeable leukocytosis and a significant thrombocytosis. This change in blood count maintains despite the initiated treatment. For this age category, the most frequent causes of leukocytosis such as infections, inflammations and allergic reactions are excluded. Also, the hematologic examination puts out a haematological malignancy.
From the patient's medical history, we summon into mind that on repeated blood count evaluations, same changes were recorded: persistent leukocytosis combined with significant thrombocytosis.
Each time, peripheral blood smears indicate poikilocytosis with ovalocytes, red blood cells in droplet, anisocytosis with microcytosis.
Several repeated abdominal ultrasounds investigations at different times, performed by different examiners do not reveal the spleen. This fact, associated with the changes in blood count and the red cells in the peripheral blood smears, raise the suspicion of a congenital asplenia.
Splenic functions' quantification methods are based on hematological, immunological and scintigraphic parameters.
The imaging methods that were performed included abdominal ultrasound and contrast-enhanced CT.
Conclusions
Following the conducted investigations, the diagnosis of isolated congenital asplenia was established.
Management is particularly important in the case of suspected infection because the patient is at risk of fulminant infections, and due to the risk of encapsulated bacteria infections, requires prophylaxis.
Keywords:
leukocytosis, thrombocytosis, congenital asplenia

77. DIFICULTĂȚI EXPLORATORII LA PACIENȚII HEMOFILICI CU INHIBITORI – SOLUȚII MODERNE DE DEPĂȘIRE A LOR

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Introducere. Dezvoltarea de inhibitori reprezintă o provocare la pacienții cu hemofilie, agenții de bypass (BPA) fiind esenţiali pentru controlul evenimentelor hemoragice. Explorarea eficacității lor este obligatorie pentru stabilirea unui regim de terapeutic salvator.
Obiectiv. Studiul nostru preliminar are scop dezvoltarea unei strategii de laborator pentru monitorizarea eficacității tratamentului cu APCC(concentrat de complex protrombinic activat) și rFVII la pacientii hemofilici cu inhibitori, prin verificarea fiabilităţii şi valorii clinice a trei teste complementare: timp de coagulare bazat pe coagulometrie, trombelastograma (TEG) și testul de generare a trombinei (TGT).
Pacienți și metoda. Evaluarea a fost realizată pe 6 pacienți cu hemofilie A severă, cu inhibitori in titru mare tratați pentru sângerări severe și episoade cu risc hemoragic ridicat (3 - chirurgie, 1 insertie cateter central, 1 hematom subdural). Următorii parametrii au fost analizaţi: APTT, PT și IP cu metoda coagulometrica; r, k, MA cu TEG; vârf și potențialul endogen de trombina (ETP) cu TGT.
Rezultate. Metoda coagulometrică a evidenţiat că numai timpul de tromboplastină a fost scurtat, cu potențial risc trombotic. TEG a dovedit r și k semnificativ prelungite, cu îmbunătățirea rapidă a valorilor lor după perfuzia de APCC sau rFVIIa. Trombelastograma a relevat valori semnificativ scăzute pentru vârf și ETP, cu o creștere la o oră după administrarea de BPA, iar la 4 ore după administrare s-a evidentiat tendința de revenire la valorile patologice.

Concluzii. Am ajuns la concluzia, că TEG şi CAT sunt testele de explorare actuale ce exprimă calitatea controlului hemostatic, reprezentând un real sprijin pentru o terapie personalizată, adaptată.
Cuvinte cheie: hemofilie, inhibitori anti-FVIII, TEG, CAT, timpi de coagulare

EXPLORATORY DIFFICULTIES IN HAEMOPHILIA PATIENTS WITH INHIBITORS - MODERN SOLUTIONS TO OVERCOME THEM

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Introduction. Development of inhibitors represents a challenging complication in patients with haemophilia and the bypassing agents (BPA) are crucial for the control of bleeding events. The laboratory evaluation of their effectiveness is mandatory for establishing a life-saving therapy regimen.
Objective. Our preliminary study aimed the development of a laboratory strategy for monitoring the treatment effectiveness of APCC and rFVII in haemophiliacs with inhibitors, by checking the reliability and clinical value of three complementary assays: clotting-time based coagulometry, thrombelastography (TEG) and thrombin generation test (TGT).
Patients and methods. The evaluation was performed on 6 patients with severe haemophilia A with high titre inhibitors treated for severe bleedings and episodes of high haemorrhagic risk (3 - surgery, 1- central catheter insertion, 1- subdural haematoma). The following parameters have been analyzed: APTT, PT and PI with coagulometry, r,k, MA with TEG and peak and endogenous thrombin potential (ETP) with TGT.
Results. The clotting-time based assay proved that only thromboplastine time was shortened, potentially signaling a thrombotic risk. TEG displayed significantly prolonged r and k, with prompt improvement of their values after infusion of ACPP or rFVIIa. Thrombelastogram revealed also significantly low values for peak and ETP, with their increase at one hour after the administration of BPA. For both agents, at 4 hours post-administration the searched parameters revealed the tendency to pathological values.
Conclusions. We concluded, that TEG and CAT are for us the only current exploratory assays, expressing the quality of haemostatic control, representing a real support for a personalized, adapted therapy.
Key words:haemophilia, anti-FVIII inhibitors, TEG, CAT, clotting time

78. A DOUA MALIGNITATE LA COPIL - EXPERIENTA UNUI SINGUR CENTRU . STUDIU INSTITUTIONAL 1990-2014, OCOLOGIE PEDIATRICA, INSTITUTUL ONCOLOGIC BUCURESTI

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Introducere: A doua malignitate este definita ca neoplasmul histologic distinct aparut concomitent sau dupa prima malignitate. Supravietuitorii unui cancer in copilarie prezinta risc crescut dedezvoltare a unei alte malignitati (aprox 3% la 20-25 de ani dupa finalizarea tratamentului pentru primul cancer, de 3-10 ori mai mare decat in populatia generala), corelat cu factori genetici si cu tratamentul primei malignitati (chimioterapie si, in special, radioterapie).
Material si metoda: Intre 1990-2014 in sectia oncopediatrie a Institutului Oncologic Bucuresti au fost tratati 1371 pacienti avand intre 10 zile-18 ani, 6 pacienti (0,43%)au prezentat a doua malignitate la 3-14 ani dupaprima malignitate.
Rezultate: A doua malignitate-tipuri histopatologice: Osteosarcom- 2 cazuri, la 7 repectiv 14 ani dupa retinoblastom unilateral respectiv bilateral la varsta mica (2 ani); Adenocarcinom tiroidian-1 caz la 3 ani dupa rabdomiosarcom pulmonar chimio-radiotratat; Carcinom scuamos lingual la 6 ani dupa leucemie acuta limfoblastica tip B; Carcinom nediferentiat ovarian-1 caz la 2 ani dupa adenocarcinom pancreatic la o pacienta cu neurofibromatoza tip 1; Leucemie acuta mieloblastica-1caz la 4 ani dupa gliom infiltrativ de de trunchi cerebral radio-chimiotratat.
Decese: 4/6 cazuri; remisiune completa la 78lunide la diagnosticul celui de al doilea cancer: 1 caz; 1 pacient se afla inca in tratament.
Concluzii: 1.Evolutia celei de-a doua malignitati este adesea nefavorabila, probabil datorita substratului genetic asociat si limitarii administrarii anumitor citostatice (doze cumulative maxime, rezistenta la citostatice). 2.Orice supravietuitor al unui cancer in copilarie trebuie monitorizat pe termen lung pentru diagnosticul precoce al unui posibil al doilea cancer.
Cuvinte cheie: a doua malignitate, copil

THE SECOND PRIMARY MALIGNANCY IN CHILDREN: A SINGLE CENTER EXPERIENCE - INSTITUTIONAL STUDY, 1990- 2014, PEDIATRIC ONCOLOGY DEPARTMENT, INSTITUTE OF ONCOLOGY BUCHAREST

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Background: The second primary malignancies (SPM) are defined as histological distinct neoplasms that present either simultaneously or after the diagnosis of an index tumor. Childhood cancer survivors have an increased risk of developing SPM (about 3% at 20 – 25 years after initial therapy, 3 – 10X risk of the general population) correlated especially with genetics and primary cancer therapy (chemotherapy and, particularly radiotherapy).
Patients and methods: 1371 patients between 10 days and 18 years old were admitted and treated in the Pediatric Oncology Department-Institute of Oncology Bucharest between 1990- 2014. 6 out ofthese (0,43%) presented the SPM 3-14 years after the primary cancer.
Results:The histological types of SPM in our study:Osteosarcoma: 2 cases, 7 and respectively 14 years after unilateral respectively bilateral retinoblastoma in young age (2years old); Thyroid adenocarcinoma-1 case, 3 years after pulmonary rhabdomiosarcoma chemotreated and radiotreated; Toungue squamos cell carcinoma- 1case, 6 years after acute lymphoblastic leukemia; Ovarian undifferential carcinoma-1 case, 2 years after pancreatic adenocarcinoma in a patient with type I neurofibrmatosis; Acute mieloid leukemia 4 years after infiltrative brain stem glioma. Deaths: 4/6 cases; complete remission at 78 months after the diagnosis of the SPM:1 case; one patient-still under oncological treatment.
Conclusions:1 The outcome of second primary malignancies is usualy unfavorable, probably because of underliying genetic disturbances and because of the limiting use of some citostatic drugs (lifetime cumulative dose, resistance). 2. Each survivor of a childhood cancer should have an long term follow-up for early diagnosis of the second malignancy.
Keywords: second primary malignancies, children

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79. INFECTIA SISTEMICA IN PATOLOGIA ONCOLOGICA PEDIATRICA

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Introducere: Chimioterapia antineoplazica este una dintre cele mai imunosupresoare terapii, determinand episoade severe de infectii sistemice in cazul copiilor in tratament oncologic. Material si metoda: Din 585 hemoculturi recoltate in 2012 in Spitalul Sf. Maria Iasi, 100 au provenit de la 61 de pacienti din cadrul Sectiei de Hemato-Oncologie Pediatrica. Rezultate si discutii: pacientii din lotul studiat urmau chimioterapie pentru: Leucemie Acuta Limfoblastica – 28; Leucemie Acuta Mieloblastica – 6; Limfom Hodgkin – 7; Limfom non-Hodgkin – 3; Nefroblastom – 3; Neuroblastom – 2; Tumori Germinale – 3; Sarcom Ewing – 2; Rabdomiosarcom – 2; Osteosarcom – 1; Tumori Cerebrale – 2; alte tipuri de tumori – 1. Din totalul hemoculturilor 10 au fost pozitive: Stafilococ auriu patogen – 6; Piocianic – 1; Acinetobacter – 1; Enterobacteriacee – 1; E. coli – 1. Toti pacientii au primit terapie antibiotica empirica cu spectru extins si ulterior tintita in functie de antibiograma iar dintre acestia 5 au decedat. Prezentam cazul pacientului A. P. A. in varsta de 13 ani, in chimioterapie pentru Leucemie Acuta Limfoblastica T cu recidiva in Sistemul Nervos Central care, pe fondul neutropeniei severe, dezvolta flebita extensiva la nivelul mainii stangi, cauzata de P. aeruginosa. Ulterior, la o luna distanta, in ciuda terapiei antibiotice extinse conform antibiogramei prezinta hidropneumotorax cu aceeasi etiologie, in absenta sterilizarii focarului de flebita necrozata. De mentionat hemoculturile ramase negative in toata aceasta perioada precum si aparitia rezistentei la Imipenem si Meropenem. Concluzii: Infectia sistemica a fost si ramane o problema majora in patologia oncologica pediatrica, cu un impact puternic in ceea ce priveste supravietuirea acestor pacienti.
Cuvinte cheie: septicemie, chimioterapie, oncologie, copil.

SYSTEMIC INFECTION IN PEDIATRIC ONCOLOGY

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Introduction: Antineoplastic chemotherapy is one of the most immunosuppressive therapies, causing systemic infection episodes in children in cancer treatment. Material and method: Of the 585 blood cultures collected in Sf. Maria Hospital in 2012, 100 were from 61 patients from Hemato – Oncology Department. Results and discutions: patients were diagnosed with Acute Lymphoblastic Leukemia - 28; Acute Myeloid Leukemia - 6; Hodgkin Lymphoma - 7; Non-Hodgkin lymphoma - 3; Nefroblastoma - 3; Neuroblastoma - 2; Germ cell tumors - 3; Ewing Sarcoma - 2; Rhabdomyosarcoma - 2; Osteosarcoma - 1; Brain tumors - 2; other tumor types - 1. Out of the total 10 were positive: Staphylococcus aureus pathogen - 6; Pseudomonas - 1; Acinetobacter - 1; Enterobacteriaceae - 1; E. coli - 1. All patients received empirical extended antibiotic therapy and subsequently targeted based on antibiogram; five of them died. We present APA patient aged 13 years in chemotherapy for relapsed Acute Lymphoblastic Leukemia T; amid severe neutropenia develops extensive phlebitis in the left hand caused by

P. aeruginosa. Subsequently, a month away, despite extensive antibiotic therapy according to antibiogram shows hidropneumotorax - same etiology, in the absence of sterilization outbreak of necrotic phlebitis. We mention repeated negative blood cultures during this period and the emergence of resistance to Imipenem and Meropenem. Conclusion: Systemic infection was and remains a major problem in pediatric oncology pathology, with a strong impact in terms of the survival of these patients.
Keywords: sepsis, chemotherapy, oncology, child.

80. DISCUTII PE MARGINEA UNUI CAZ DE HEPATOBLASTOM

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In ciuda faptului ca hepatoblastomului este cea mai comuna tumora hepatica, reprezinta o patologie rară descoperită la pacienții pediatrici. Aceasta reprezintă aproximativ 1% din totalul cancerelor pediatrice.
Autorii prezinta cazul unui copil in varsta de 2 ani cu o masa abdominala, care a fost internat in Clinica Pediatrie I Tg-Mures-Compartiment Hematooncologie. Ecografia abdominala si CT-abdominal au evidentiat o tumora solida hepatica. Valoarea serica a LDH-ului si alfa- fetoproteina au fost crescute. Pacientul prezenta trombocitoza si biopsia hepatica a fost efectuata. Examinarea histopatologica a evidentiat un hepatoblastom embrionar. Odata ce diagnosticul a fost confirmat, s-a initiat chimioterapie, urmata de interventie chirurgicala in doua etape: ligatura ramurei drepte a venei porte, urmat de rezectia tumorii.
Tratamentul primar al hepatoblastomului este rezectia chirurgicala, iar combinația intre chimioterapie si tehnici chirurgicale s-a dovedit a fi o bună opțiune de tratament.
Cuvinte cheie: hepatoblastom, copil, alfa-fetoproteina

DISCUSSION ON A CASE OF HEPATOBLASTOMA

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Despite the fact that hepatoblastoma is the most common hepatic tumor, it is a rare pathology discovered in pediatric patients. It represents about 1% of total pediatric cancers. The authors present the case of a two-years old patient with an abdominal mass who has admitted in the Pediatric Clinic I Tg-Mures-Hematooncology Department. Ultrasound and CT scan demonstrated a solid hepatic tumor. Serum LDH and alpha fetoprotein level was increased. He presented thrombocytosis and liver biopsy was performed. The microscopic examination demonstrated the histological aspect of a embryonic epithelial hepatoblastoma. Once the diagnosis is confirmed, chemotherapy is initiated, followed by surgery in two steps: the right portal branch ligation and then tumor resection was performed.
Hepatic tumor at a child under 3 years old correlated with elevated serum alpha fetoprotein and thrombocytosis are almost patognomonic for hepatoblastoma. The primary treatment is surgical resection, and the combination of chemotherapeutic regimes and surgical techniques has shown to be a good treatment option.
Key words: hepatoblastoma, child, alpha fetoprotein

81. NEUTROPENIA CRONICA - PARTICULARITATI CLINICO-EVOLUTIVE

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Neutropenia cronica la copil este o cauza frecventa a infectiilor severe. Pentru tarile in curs de dezvoltare exista studii putine privind aceasta patologie.
Material si metode: Au fost evaluati pacientii(0-18 ani) cu neutropenie cronica cu o durata de minim 6 luni. S-au exclus pacientii cu pancitopenie ,cei care au primit anterior chimioterapie si cei cu boli maligne. Studiul a fost retrospectiv pe perioada ianuarie 2002-decembrie 2014.
Rezultate: Lotul a cuprins 35 de pacienti cu neutropenie cronica (15 fete, 20 baieti): 6 cu neutropenie congenitala severa, 19 cu neutropenie autoimuna si 10 cu neutropenie idiopatica. Durata medie de urmarire a fost de 5,5 ani(1-11,9). G-CSF s-a utilizat la 16 din 35 pacienti, respectiv la toti pacientii cu neutropenie severa congenitala, 9 din 19 pacienti cu neutropenie autoimuna si 1 din 10 cu neutropenie idiopatica. S-au inregistrat 120 episoade infectioase care au necesitat spitalizare: 11 amenintatoare de viata; 92 la pacientii cu neutropenie congenitala severa. Caracterizarea microbiologica a fost posibila in 23 de cazuri. Cele mai frecvente bacterii au fost Staphylococcus aureus(9 cazuri), *Escherichia coli(*5 cazuri), *Klebsiella pneumoniae*(2 cazuri), *Pseudomonas aeruginosa*(2 cazuri*), Enterococcus*(2 cazuri). Infectiile la nivelul cavitatii bucale(17%), pneumonia(16%), otitele(15%), infectiile cutanate(15%) au fost cele mai frecvente. Durata medie de spitalizare cea mai indelungata a fost pentru pacientii cu neutropenie congenitala severa(14,3 zile).

Concluzii: Cel mai mare numar de infectii a fost evidentiat la pacientii cu neutropenie severa congenitala, iar cavitatea orala a fost sediul cel mai implicat. Desi G-CSF s-a utilizat doar on demand, nu s-a inregistrat nici un deces prin sepsis.
Cuvinte cheie: neutropenie cronica, G-CSF, infectii

CHRONIC NEUTROPENIA - CLINICAL AND EVOLUTIVE ASPECTS

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Chronic childhood neutropenia is a frequent cause of severe infections. There are few studies conducted in the developmental phase countries concerning this pathology.
Material and methods: Patients (0-18 year) with chronic neutropenia ongoing for minimum 6 months were evaluated. Patients with pancytopenia, with anterior chemotherapic treatment and those with malignancies were excluded. We reviewed the charts of all such patients seen at our center from January 2002 to December 2014.
Results: A total of 35 patients with chronic neutropenia (15 f, 20 m): 6 with SCN, 19 with AN and 10 with IN. the average follow-up period was 5,5 years (1-11,9). G-CSF was used on 16 out of 35 patients, respectively on all patients with SCN, 9 out of 19 with AN and 1 out of 10 with IN. 120 infectious episodes that needed hospitalization were recorded: 11 life threatening; 92 patients with SCN. Microbiological identification was possible for 23 cases. The most frequently implied bacteria were Staphylococcus aureus(9 cases), *Escherichia coli(*5 cases), *Klebsiella pneumoniae*(2 cases), *Pseudomonas aeruginosa*(2 cases*), Enterococcus*(2 cases). The most frequent were oral cavity infections 17%, pneumonia 16%, otitis 15%, cutaneous infections 15%. The average hospitalization period was longer for patients with SCN (14,3 days).
Conclusion: Patients with SCN presented most infectious episodes. Oral cavity was the most frequent implied site. Although G-CSF was only used on demand, there was no death caused by sepsis.
Key word: Chronic neutropenia, G-CSF, infections

82. BOALA HODGKIN SI SINDROMUL NEFROTIC LA COPII

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Introducere:
Asocierea sindromului nefrotic in Boala Hodgkin este cunoscuta din anul 1992. Frecventa acestei asocieri este de 1%.
Material si metoda:
In perioada 1997-2015 au fost tratati 107 copii cu Boala Hodgkin in sectia de Oncopediatrie SCUC "M.S. Curie". Dintre acestia 2 cazuri (2%) au avut asociat sindromul nefrotic.
In primul caz (1997), baiat in varsta de 7 ani, diagnosticul de sindrom nefrotic a precedat diagnosticul de B. Hodgkin cu 3 ani desi imagistic prezenta o formatiune tumorala localizata in mediastinul anterior de 3 cm.
In perioada 1995-1997 a fost efectuata corticoterapie dar sindromul nefrotic recadea rapid la intreruperea tratamentului.
Diagnosticul de B.Hodgkin a fost stabilit dupa rezectia unei formatiuni tumorale mediastinale de 15cm (1997) iar tratamentul chimioterapic (cure MOPP/ABVD) si radioterapic au vindecat ambele afectiuni.
Al doilea caz in anul 2015, baiat in varsta de 9 ani, sindromul nefrotic a debutat cu 3-4 luni inainte de stabilirea diagnosticului de Boala Hodgkin fiind o forma rezistenta la tratament.
Si in acest caz tratamentul limfomului (protocol EURONET PHL C1 – cura OEPA) a determinat disparitia rapida (dupa prima cura) a manifestarilor sindromului nefrotic.
Concluzii:
Boala Hodgkin este rar asociata cu sindrom nefrotic (2%). In aceste cazuri sindromul nefrotic raspunde complet numai la tratamentul bolii de baza (B. Hodgkin).
In caz de recadere a bolii Hodgkin la acesti pacienti reapare frecvent si sindromul nefrotic.
Este posibil ca tulburarile imunologice din Boala Hodgkin sa joace un rol in patogeneza acestei asocieri – sindrom nefrotic si Boala Hodgkin.
Cuv.cheie: Boala Hodgkin, sindrom nefrotic.

HODGKIN LYMPHOMA AND NEPHROTIC SYNDROME IN CHILDREN. REPORT OF TWO CASES

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Introduction:
An association between nephrotic syndrome and Hodgkin lymphoma was described for the first time in 1992. The incidence of nephrotic syndrome in Hodgkin lymphoma is less than 1%.
Material and method:

In the period between 1997-2015 in Oncopediatrics Department of "M.S.Curie" Hospital were treated 107 children with Hodgkin lymphoma. Only 2 cases associated nephrotic syndrome.
Case 1 (1997) – a 7 year old boy, the nephrotic syndrome predated the diagnosis of lymphoma by 3 years, even though the CT showed a tumor localised in anterior mediastinum (3cm).
Between 1995-1997 he received immunosuppressants, but the nephrotic sydrome relapsed immediatly after the disruption of the treatment.
The diagnosis of Hodgkin lymphoma was established after the resection of the 15 cm mediastinal tumor , than he received chemoterapy (protocol MOPP/ABVD), followed by involved field radiation, remission of his lymphoma and nephropathy was achieved.
Case 2 (2015) – a 5 year old boy , the nephrotic syndrome predated the diagnosis of lymphoma by 3- 4 months but the nephrotic syndrome relapsed frequently.
In this case the chemoterapy (protocol EURONET PHL C1 – OEPA) determined a fast remission of the nephrotic syndrome.
Conclusions:
The association of nephrotic syndrome and Hodgkin disease is very rare, in this cases the nephrotic syndrome responds only at the treatament of the Hodgkin lymphoma.
The patient with the return of Hodgkin lymphoma have also the relapsing of the nephrotic syndrome.
The occurence of nephrotic changes as manifestation of HL, suggests that some immunological abnormalities may play a role in the pathogenesis of this association.
Key words: Hodgkin lymphoma, nephrotic syndrome

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83. CHISTUL OVARIAN DERMOID – PREZENTARE DE CAZ

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Introducere:Chistul dermoid sau teratomul ovarian este o formatiune chistica plina cu tesut: piele, glande sudoripare, par, os, dinti, unghii, care isi are originea in celule de origine embrionara ramase in tesutul ovarian. Chistul dermoid reprezinta 20-25% din tumorile ovariene si este bilateral in 10-13% din cazuri. La copii si adolescenti este cea mai frecventa tumora , reprezentand peste jumatate din neoplasmele ovariene la femeile mai tinere de 20 ani. Majoritatea sunt asimptomatice fiind descoperite intamplator. Datorita complicatiilor ce pot aparea:torsiunea, ruptura spontana, peritonita chimica se impune tratamentul chirurgical in momentul diagnosticarii.
Metoda: Autorii descriu cazul unei adolescente de 18 ani care se prezinta pentru distensie abdominala marcata, constipatie, dureri in etajul abdominal inferior. Investigatiile paraclinice hemoleucograma, reactantii de faza acuta, AFP si antigenul CA-125 au fost in limite normale. Testul de sarcina a fost negativ. Examenul ecografic a evidentiat o formatiune chistica bine delimitata neomogena, uter laterodeviat stanga de formatiune, ovarul stang de aspect normal. Se stabileste diagnosticul de chist ovarian drept si se decide interventie chirurgicala laparoscopica. Intraoperator dupa incizia chistului se extrag fragmente osoase, tesut gras si fire de par orierntand diagnosticul catre un chist dermoid, care a fost confirmat prin examen histopatologic.
Concluzie: dimensiunile chistului au facut necesara ovarectomia totala. Postoperator evolutia a fost favorabila.
Cuvinte cheie: chist dermoid, teratom ovarian, adolescente

OVARIAN DERMOID CYST – CASE REPORT

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Introduction: Dermoid cyst or ovarian teratoma is a cystic formation filled with tissue: skin, sweat glands, hair, bone, teeth, nails, which originates in embryonic cells of remaining ovarian tissue. Dermoid cyst represents 20-25% of ovarian tumors and it is bilateral in 10-13% of cases. In children and adolescents is the most common tumor, representing more than half of ovarian malignancies in women younger than 20 years old. Most of them are asymptomatic being discovered by chance. Due to complications that may occur: torsion, spontaneous rupture, chemical peritonitis, surgery is required at the moment of diagnosis.
Method: The authors describe the case of an 18 years old teenager who was hospitalised for marked abdominal distention, constipation, lower abdominal pain. Laboratory investigations: complete blood count(CBC), acute phase reactants, AFP and CA-125 antigen were within normal limits. Pregnancy test was negative. Ultrasound examination revealed a well defined cystic heterogeneous pattern, uterus deflected the left side of the pattern, left ovary of normal aspect. Right ovarian cyst

diagnosis is established and a laparoscopic surgery is performed. During surgery, after cyst incision, bone fragments, hair and fatty tissue were extracted,which orientated the diagnosis to a dermoid cyst, which was confirmed by histopathology.
Conclusion: cyst size made necessary total ovariectomy. Postoperatory evolution was favorable.
Keywords: Dermoid cyst ,ovarian teratoma, teenager

84. EVALUAREA RASPUNSULUI LA TERAPIA IMUNOSUPRESOARE CU GLOBULINA ANTITIMOCITARA DERIVATE DIN PROTEINA DE CAL VERSUS PROTEINA DE IEPURE

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Introducere. Anemia aplastica (AA) dobandita este o afectiune rara, dar potential fatala, care poate fi diagnosticata la pacientii pediatrici sau adulti. Diferentierea formelor dobandite de AA de cele constitutionale sau de sindromaele mielodisplazice hipocelulare este esentiala pentru managementul correct al pacientilor. Terapia standard a pacientilor cu forme severe sau foarte severe de AA este reprezentata de transplantul de celule stem hematopoietice in cazul identificarii unui donator familial compatibil sau de terapia imunosupresoare in cazul in care nu exista un donator familial compatibil. Terapia imunosupresoare presupune asocierea de globulina antitimocitara (ATG), ciclosporina si metilprednisolon. Produsele comerciale care contin ATG pot fi derivate din ser de cal sau ser de iepure, iar studii recente demonstreaza raspunsuri diferite la aceste medicamente, astfel incat ghidurile terapeutice actuale recomanda utilizarea initiala de ATG derivat din cal (ATGAM).
Scop. Evaluarea raspunsului hematologic si analiza complicatiilor secundare admninistrarii ATG derivate din ser de cal (ATGAM - Pfizer), Lymphoglobulin (Genzyme) comparativ cu ATG derivate din ser de iepure (Thymoglobulin (Sanofi) sau ATG –Fresenius).
Pacienti si metode. Am realizat un studiu retrospectiv, observational, care a inclus toti pacientii cu AA severa sau foarte severa internati in Clinica de pediatrie Fundeni in perioada 2000-2015 la care s-a administrat orice tip de produs ATG. Diagnosticul de AA s-a efectuat pe baza analizei datelor clinice, hematologice (hemoleucograma, reticulocite, indici eritrocitari, examen histopatologic al biopsiei osoase), biochimice, examen citogenetic standard, teste virusologice (HIV, VHB, VHC, CMV, EBV). Formele de AA severa au fost definite de celularitate medulara <25% + $\geq$ 2 din urmatoarele: neutrofile < 0,5 x10^9/L, trombocite < 20 x 10^9/L, Rt < 40x10^9/L, iar formele foarte severe de aceleasi criteria cu exceptia numarului de neutrofile care este <0,2 x10^9/L. Imunosupresia (IS) s-a realizat cu ciclosporina 5mg/kgC/zi, metilprednisolon si ATG in doza specifica functie de produsul utilizat. Raspunsul la IS a fost apreciat prin a) monitorizarea parametrilor hematologici (neutrofile, trombocite, reticulocite), b) evaluarea necesarului transfuzional (nr. unitati de MEr si MTr) si c) evaluarea celularitatii medulare prin biopsie la 3 l, 6 l de la administrare. A fost evaluat intervalul de timp pana la obtinerea raspunsului hematologic sau pana la terapia de linia a 2a.
Rezultate. In perioada 2000-2015 in Clinica de Pediatrie a ICF au fost diagnosticati 62 de copii cu anemii aplastice: 16 forme constitutionale (Anemie Fanconi 8, Anemie Blackfan Diamond 4, Discheratoza congenitala 2, forme neincadrabile 2) si 47 forme dobandite. Zece pacienti (5M/5F) au primit ATG in cadrul programului de IS: Lymphoglobulina 3, ATGAM 3, Thymoglobulina 3, ATG-Fresenius 1. Raspunsul hematologic complet s-a inregistrat la 4 pacienti (ATGAM 2, Lymphoglobulin 2), raspuns hematologic incomplet + independent transfuzionala s-a inregistrat la 3 pacienti (ATGAM 1, Thymoglobulin 2), absenta raspunsului hematologic s-a constatat la 3 pacienti (Thymoglobulin 1, Lymphoglobulin 1, ATG-Fresenius 1). Doi dintre pacientii fara raspuns hematologic au efectuat transplant de celule stem hematopoietice si sunt in remisiune hematologica completa, 1 pacient a decedat ca urmare a unui episod infectios sever. Raspunsul hematologic complet s-a observat la pacienti care au primit ATG in primele 3 luni de la diagnostic.
Discutii/Concluzii. Preparatele care contin ATG, desi au aceeasi activitate T citotoxica, sunt foarte diferite in ceea ce priveste activitatea anti NK. Raspunsul la administrarea ATG derivate din proteina de cal este mai rapid si apare la un procent mai mare de pacienti.

EVALUATION OF RESPONSE TO IMMUNOSUPPRESSIVE THERAPY WITH ANTITHYMOCYTE GLOBULIN DERIVED FROM HORSE PROTEIN VERSUS RABBIT PROTEIN

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Introduction. Acquired aplastic anemia (AA) is a rare but potentially fatal condition that can be diagnosed in pediatric patients or adults. Differentiation between acquired and constitutional AA and hypo cellular myelodysplastic syndromes (MDS) is essential for correct management of patients. Standard therapy in patients with severe or very severe forms of AA is represented by hematopoietic stem cell transplantation (HSCT), when there is a compatible family donor or immunosuppressive (IS) therapy where there is no compatible family donor. Immunosuppressive therapy is represented by the combination of antithymocyte globulin (ATG), cyclosporine and methylprednisolone. Commercial products containing ATG can be derived from horse serum or rabbit serum, and recent studies show different responses to these drugs, so current treatment guidelines recommend initial use of horse-derived ATG (ATGAM).

Purpose. Hematologic response assessment and analysis of secondary complications after administration of ATG derived from horse serum (ATGAM - Pfizer), Lymphoglobulin (Genzyme) compared with ATG derived from rabbit serum (Thymoglobulin (Sanofi) or Fresenius ATG).

Patients and methods. We conducted a retrospective, observational study, which included all patients with severe or very severe AA hospitalized in Fundeni Pediatric Clinic in the period 2000-2015 who received any type of ATG product. AA diagnosis was based on clinical data analysis, hematological (blood count, reticulocytes, red cell indices, bone marrow biopsy histopathology), biochemical, cytogenetic examination, standard viruses tests (HIV, HBV, HCV, CMV, EBV). AA severe forms were defined by bone marrow cellularity <25% + ≥ 2 of the following: neutrophil count <0.5 x109 / L, platelets <20 x 109 / L, R <40x109 / L and severe forms with the same criteria but neutrophil count <0.2 x109 / L. Immunosuppression (IS) was with cyclosporine 5mg/kg/day, methylprednisolone and ATG (dose depending on the product specifications). The answer to IS was assessed by a) monitoring of hematological parameters (neutrophils, platelets, reticulocytes), b) evaluating transfusion requirements (no. of blood units) and c) evaluating bone marrow biopsy cellularity at 3 mo and 6 mo from administration. We evaluated the time until hematological response was achieved or until second-line therapy.

Results. During 2000-2015 in the Pediatric Clinic of Fundeni Clinical Institute 62 children were diagnosed with aplastic anemia: 16 constitutional forms (8 – Fanconi Anemia, 4 – Diamond Blackfan Anemia, 4 – Diskeratosis congenita, 2 – unclassified BMF syndromes) and 47 acquired AA. Ten patients (5M/5F) received ATG: Lymphoglobulin – 3 cases, ATGAM – 3 cases, Thymoglobulin – 3 cases, ATG-Fresenius – 1 case. Complete hematologic response was seen in 4 patients (ATGAM – 2 cases, Lymphoglobulin – 2 cases), incomplete hematological response + no transfusion requirement – occurred in 3 patients (ATGAM – 1 case, Thymoglobulin – 2 cases), the absence of hematologic response was found in 3 patients (Thymoglobulin – 1 case, Lymphoglobulin – 1 case, ATG – Fresenius – 1 case). Two of the patients without hematologic response performed hematopoietic stem cell transplantation and are now in complete hematologic remission, one patient died following a severe infectious episode. Complete hematologic response was observed in patients who received ATG in the first 3 months after diagnosis.

Discussion/Conclusion**.** Products that contain ATG, though having same cytotoxic T activity, are very different in terms of anti NK. The answer to ATG derived from horse protein is faster and occurs at a higher percentage of patients.

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85. FACTORI PREDISPOZANTI PENTRU INFECTIA CU POLYOMAVIRUSURI LA PACIENTII CU ALLOGREFA

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Introducere

Infectiile virale reprezinta o complicatie majora a transplantului de celule stem hematopoietice, cu o incidenta mai redusa comparativ cu infectiile fungice sau bacteriene, dar frecvent cu evolutie mai grava si mortalitate ridicata. În cazul pacientilor transplantati, replicarea virala si progresia catre boala sunt determinate de factori specifici, dependenti de pacient, de donator si de virus.

Reactivarea polyomavirusurilor (BKV sau JCV) se asociaza cu cistita hemoragica; infectia BKV este notata la 13% dintre copii posttransplant de celule stem hematopoietice. Cistita BKV apare tipic la momentul grefarii si este asociata cu valori crescute si persistente ale viruriei BKV. BKV este frecvent identificat si in sange, sugerand ca inflamatia vezicii urinare determina invazia sanguina.

Obiective

Am realizat un studiu retrospectiv pentru a analiza: a) incidenta si evolutia infectiilor virale post-transplant de celule stem hematopoietice (CSH) la pacientii pediatrici la care s-a realizat monitorizarea prin tehnici PCR; b) interrelatia dintre infectia virala si statusul imunosupresiei necesare pentru controlul reactiei de grefa contra gazda (GVHD – graft versus host disease).

Material si metode

Lotul de studiu a inclus 38 copii care au efectuat allotransplant de CSH in Centrul de Transplant Medular al IC Fundeni. Studiul realizat este retrospectiv.

Compatibilitatea HLA intre donator si primitor a fost demonstrata la nivel allelic pentru locii HLA de clasa I si II. Inainte de transplant s-a realizat analiza ELISA pentru anticorpi specifici IgG si IgM pentru CMV si EBV atat la donator, cat si la primitor. Profilaxia infectiilor virale s-a realizat cu Aciclovir. Detectarea infectiilor virale s-a realizat prin teste RT-PCR pentru CMV, EBV, HSV1, HHV6, HHV8, ADV, VZV si teste Nested PCR pentru JCV si BKV.

Rezultate

Testarea virala a permis identificarea a 6 cazuri de infectie cu poliomavirusuri.

Cistita hemoragica cu BKV a fost identificata la 5 pacienti cu allotransplant de celule stem hematopoietice. Testele Nested PCR au fost pozitive in ser si urina. Evolutia cistitei a variat de la cateva zile pana la 2 luni si a raspuns doar la reducerea tratamentului imunosupresor. Pacientii au dezvoltat concomitent si infectie CMV.

Cistita hemoragica cu JCV a fost diagnosticata la un pacient cu allotransplant medular pentru beta talasemie majora concomitent cu boala CMV (hepatita si encefalita). Testele Nested PCR

au fost pozitive atat in sange, cat si in urina, iar ecografia vezicala a aratat ingrosarea semnificativa a mucoasei vezicale. Evolutia cistitei a fost indelungata (6 saptamani), a necesitat tratament suportiv si antialgic intens si a raspuns doar la reducerea tratamentului imunosupresor.

Concluzii si discutii

Monitorizarea polyomavirusurilor in urina este importanta pentru stabilirea diagnosticului etiologic al cistitei hemoragice posttransplant, dar abordarea terapeutica a acestei complicatii ramane neschimbata, atata timp cat nu exista medicamente cu actiune specifi ca anti BKV sau JCV.

Incidenta redusa a infectiei cu polyomavirusuri nu justifica un program de monitorizare, doar diagnostic in momentul aparitiei febrei, disuriei, hematuriei sau urmarirea simptomelor sugestive pentru infectie virala.

Toti pacientii la care s-a identificat cistita cu polyomavirusuri au prezentat si coinfectie CMV.

Infectiile cu polyomavirusuri au aparut la toti pacientii in contextul imunosupresiei combinate pentru GVHD.

PREDISPOSING FACTORS TO POLYOMAVIRUSES INFECTION IN PATIENTS WITH ALLOGRAFT

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Introduction

Viral infections are a major complication of hematopoietic stem cell transplantation (HSCT) with reduced incidence compared to bacterial or fungal infections, but often with more severe evolution and mortality. In transplanted patients, viral replication and progression of the disease are caused by specific factors, depending on patient, donor and virus.

Polyomaviruses reactivation(BKV and JCV) is associated with hemorrhagic cystitis; BKV infection is noted in 13% of children post HSCT. BKV cystitis typically occurs at the time of engraftment and is associated with elevated and persistent presence of viral DNA in urine. BKV is also frequently found in the blood, suggesting that bladder inflammation leads to blood invasion.

Objectives

We performed a retrospective study to analyze: a) the incidence and evolution of viral infections post HSCT in pediatric patients who were monitored using PCR techniques; b) the interrelation between viral infection and immunosuppression status needed to control the reaction of graft versus host disease (GVHD - graft versus host disease).

Material and Methods

The study group included 38 children who underwent allo-HSCT in the Fundeni Clinical Institute BMT Center. The study is retrospective.

HLA compatibility between donor and recipient was demonstrated at allelic loci HLA class I and II. Before transplant CMV and EBV IgG and IgM antibodies were performed for both the donor and the recipient. Viral infection prophylaxis was done with acyclovir. Detection of viral infections was done by RT-PCR tests for CMV, EBV, HSV1, HHV6, HHV8, ADV, VZV and Nested PCR assays for BKV and JCV.

Results

Viral testing lead to identification of six polyomaviruses infections.

Hemorrhagic cystitis with BKV was identified in 5 patients with allo-HSCT. Nested PCR tests were positive in serum and urine. Evolution of cystitis varied from several days to two months and responded only to reducing immunosuppressive therapy. The patients developed CMV infection concomitantly.

JCV hemorrhagic cystitis was diagnosed in a patient with major beta thalassemia who underwent allo-HSCT, concomitantly with CMV disease (hepatitis and encephalitis). Nested PCR tests were positive in both blood and urine and bladder ultrasound showed significant thickening of the lining of the bladder. Evolution of cystitis was long (6 weeks), requiring intensive supportive care and pain reliever and responded only to reducing immunosuppressive therapy.

Conclusions and discussion

Monitoring polyomaviruses in urine is important for diagnosis of hemorrhagic cystitis post-transplantation, but the management of this complication remains unchanged as long as there are no specific drugs for BKV or JCV.

The reduced incidence of polyomaviruses infection does not justify a monitoring program, just diagnosis when symptoms occur: fever, dysuria, hematuria or monitoring symptoms suggestive for viral infection.

All patients who had cystitis with polyomaviruses had CMV coinfection. Polyomaviruses infections occurred in all patients in the context of combined immunosuppression for GVHD.

GENETICĂ / GENETICS

86. BOALA LIZOZOMALA CU PROGNOSTIC SEVER - GM1 GANGLIOZIDOZA

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GM1 gangliozidoza este o boala autozomal recesiva rara, cauzata de mutații ale genei GLB1, ce codeaza beta-galactozidaza lizozomala.

Scopul lucrarii este prezentarea caracteristicilor epidemiologice si clinico-evolutive pentru trei pacienti avand acest diagnostic, internati în Spitalul Clinic de Urgenta pentru Copii ”MS Curie”, in perioada 2004-2015. Prevalenta bolii este raportata la un numarul total de internari, din cei 11 ani, adică 300 000. Sunt luați in discuție mai mulți parametri clinico-biologici.

Rezultate: Toti 3 pacientii au fost de origine romana. Diagnosticul de GM1-gangliozidoza a fost confirmat enzimatic la 3/3 cazuri, la varstele de 9, 8, respectiv 9 luni. Diagnosticul molecular s-a efectuat la 1/3 cazuri. Un caz are un precedent in fratrie. Tabloul clinic a inclus edeme (3/3 cazuri), anasarca (2/3), cardiomiopatie hipertrofica (2/3), hepatosplenomegalie (3/3), falimentul cresterii (2/3), atrofie optica (2/3), retard de dezvoltare si convulsii (3/3 cazuri). Evoluția a fost grevată de complicatii respiratorii – bronhopnemonie (3/3), insuficienta cardiaca acuta (1/3) sau cronica (1/3), anemie (2/3), trombocitopenie (2/3) si sepsis (3/3). Terapia a presupus alimentație speciala pe gastrostoma sau SNG, abord venos central, tonicardiac (2/3 cazuri), diuretic (3/3 cazuri), sedare pentru convulsii (2/3 cazuri) + IOT (1 caz), antibiotice de rezervă (carbapenemi, vancomicina) și transfuzii (1/3 cazuri). Decesele s-au produs prin complicatii respiratorii, la varste cuprinse intre 10 luni- 1 an.

Concluzii: Prevalenta anuala a GM1 gangliozidozei a fost de 1/100.000 internari. Suferința severa respiratorie, cardiaca si neurologica, precum si complicațiile septice necesită facilitati de terapie intensiva. Prognosticul este rezervat, in absența terapiei de substitutie enzimatica.

GM1- GANGLIOSIDOSIS – A LYSOSOMAL STORAGE DISEASE WITH SEVERE PROGNOSIS

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GM1-gangliosidosis is a rare autosomal recessive lysosomal disease, caused by mutations in GLB1 gene, encoding the beta-galactosidase. Our aim is to present the epidemic and clinical aspects for three patients diagnosed with GM1-gangliosidosis, admitted in the Clinical Emergency Hospital for Children ”MS Curie”, during the 2004-2015 period. Prevalence of the disease is reported to the total number of hospitalized children, meaning 300 000. Different clinical-biological parameters were assesed.

Results: All 3 patients were from Romanian origin. The specific diagnosis was made upon enzymatic assay of serum beta-galactosidase, for all 3/3 cases, at the age of 9, 8, and 9 months. Molecular diagnosis was performed in 1/3 cases. One patient is the second child affected in the family. Clinical picture included: edema (3/3 cases), hypertrophic cardiomyopathy (2/3), hepatosplenomegaly (3/3), failure to thrive (2/3), optic atrophy (2/3), developmental delay and seizures (3/3 cases). Evolution was marked by complications such as: bronchopnemonia (3/3), acute (1/3) or cronic (1/3) heart failure, anemia (2/3), thrombocytopenia (2/3) and sepsis (3/3). Therapy required special nutrition given by gastrostomy or nasogastric tube, central venous catheter, cardiotonic agents (2/3 cases), diuretics (3/3 cases), sedation for seizures (2/3 cases) + OTI (1 case), reserve antibiotics (carbapenems, vancomycin) and transfusions (1/3 cases). Death occurred due to respiratory complications, at the age of 10-12 months.

Conclusions: The annual prevalence of GM1-gangliosidosis was 1/100.000 hospitalized patients. The severe respiratory distress, cardiac and neurologic impairment require intensive care facilities. Prognosis is reserved, in the absence of a specific enzymatic replacement therapy.

Key-words: GM1 gangliosidosis, diagnosis, evolution

87. BOALA WERDNIG-HOFFMAN ÎN FRATRIE PREZENTARE DE CAZ

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Boala Werdnig-Hoffman, amiotrofia spinală tip I (BWH), este o boală neuromusculară progresivă, rară, caracterizată prin degenerescenţa neuronilor motori și determinată de deleții sau anomalii ale genei SMN1.

Autorii prezintă cazul unei paciente în vârstă de 5 luni și 2 saptamani cu BWH cu debut precoce, cu tulburări și complicații respiratorii și digestive secundare hipotoniei musculare și abolirii reflexelor – cu evoluție rapidă spre deces. Pacienta provine din familie cu istoric cunoscut de BWH (sora mai mare).

Cuvinte-cheie: Boala Werdnig-Hoffmanm tip I, debut precoce, evoluție letală.

THE WERDNIG-HOFFMAN DISEASE IN A BROTHERHOOD - CASE REPORT

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The Werdnig-Hoffman disease, spinal muscular atrophy type I (SMA-I), is a rare, progressive neuro-muscular disease characterized by degeneration of the motor neurons and induced by deletions or other abnormalities of the SMN1 gene.
The authors report a 5months 2 weeks old female patient with early onset SMA-I, who manifests respiratory and digestive tract alterations second to muscular hypotony and reflex abolition – with rapid lethal progression. The patient comes from a family with a known history of SMA-1 (an elder sister).
Key-words: Werdnig-Hoffman disease type I, early onset, lethal progression.

88. PARTICULARITATI CLINICE SI DE DIAGNOSTIC LA UN PACIENT CU SINDROM V.A.T.E.R.

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Sindromul V.A.T.E.R. sau V.A.C.T.E.R.L. este un acronim folosit pentru a descrie o asociere non-aleatoare de malformatii congenitale. V.- anomalii vertebrale, A.- agenezie anala, C.- anomalii cardiace, T.-E.- fistula traheo-esofagiana, R.-anomalii renale, L.- anomalii ale structurii membrelor.
Pentru stabilirea diagnosticului este necesara asocierea a cel putin 3 anomalii, indiferent de gradul de severitate al acestora.
Lucrarea de fata urmareste cazul unui sugar, in evidenta Spitalului Clinic de Urgenta “Grigore Alexandrescu” inca de la nastere, diagnosticat la varsta de 6 luni cu sindromul V.A.C.T.E.R.L
Din antecedentele personale fiziologice ale pacientului mentionam ca acesta provine dintr-o sarcina nedispensarizata, cu nastere spontana, la termen, fara antecedente heredo-colaterale semnificative.
Din antecedentele patologice ale acestuia retinem ca la 24 de ore de la nastere este adus in clinica noastra in stare generala grava, diagnosticat cu sindrom plurimalformativ osos, agenezie anala, pentru care se intervine chirurgical in urgenta si se instituie colostoma dreapta, si testicul stang necoborat in scrot.
Pacientul este investigat imagistic si se identifica: multiple malformatii scheletice: hemivertebra C7, multiple situsuri de fuziune vertebrala incompleta la nivel T4-T8-13, 13 arcuri costale drepte, 11 arcuri costale stangi, hipoplazie costala stanga C III stang, defect septal ventricular, foramen ovale permeabil, rinichi drept malrotat, ectopic, reflux vezico-ureteral bilateral, si malformatie police stang, intrunind astfel 5 dintre cele 6 criterii de diagnostic pentru sindromul V.A.C.T.E.R.L.
Particular acestui caz este prezenta a 5 din cele 6 criterii de diagnostic, cat si absenta fistulei traheo-esofagiene; una dintre cele mai frecvent intalnite malformatii in asocierea V.A.C.T.E.R.L.

CLINICAL AND DIAGNOSTIC PARTICULARITIES IN A CASE OF VATER SYNDROME

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V.A.T.E.R. syndrome or V.A.C.T.E.R.L. is an acronym used to describe a non-random association of birth defects. V.- vertebral anomalies, A.-anal agenesis, C.- cardiac abnormalities, T.-E.-tracheo-esophageal fistula , R.-renal abnormalities, L.- structure abnormalities of the limbs.
For a positive diagnosis a combination of at least three abnormalities is necessary, regardless of their degree of severity.
The following presentation is centered on the case of an infant in the evidence of the Emergency Hospital for Children "Grigore Alexandrescu" since birth, diagnosed with VACTERL syndrome at the age of 6 months.
From the patient's personal history we mention that it originates from an uneventful, uninvestigated pregnancy, with a spontaneous delivery, with no significant familial history.
The patient’s medical history reveals that 24 hours after it’s birth, he was brought into our clinic with severe general state, diagnosed with skeletal plurimalformative syndrome, left undescended testicle and anal agenesis, for which immediate surgical intervention, followed by a colostomy establishment was needed.
Further imaging investigations showed: multiple skeletal malformations: C7 hemivertebra, incomplete spinal fusion in multiple sites at T4-T8-13, 13 rib arches on the right side, and 11 rib arches on the left side, hypoplastic left C III rib, ventricular septal defect, permeable foramen ovale, malrotated, ectopic right kidney, vesicoureteral bilateral reflux and left police malformation, thus meeting the 5 out of 6 VACTERL syndrome diagnostic criteria
Particular in this case is the presence of 5 out of 6 diagnostic criteria and the absence of tracheo-esophageal fistula; one of the most common malformations in VACTERL association.

89. SINDROM HOLT-ORAM-DEPISTATRE TARDIVĂ-PREZENTARE DE CAZ

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Scop- de a prezenta un caz depistat tardiv la vârsta de 7 ani cu ocazia internării pentru o exacerbare a astmului bronşic.
Prezentare de caz-PG, sex masculin, 7 ani, se prezintă pentru evaluarea unui episod bronhoobstructiv formă medie. Examenul clinic depistează un copil cu deficit staturoponderal, cicatrice la nivelul palatului post cheilognatopaptoschizis operat, scolioză toracală dextroconvexă, cord ritmic, SS grad IV/6, ectrodactilie mână dreaptă-absenţa degetului I, pectus excavatum. Traseul EKG este nemodificat, iar echografia cardiacă Doppler color evidentiază defect septal atrial mic, prolaps de valvă mitrală cu insuficienţă mitrală uşoară. Radiologic se depistează o sindactilie de bază metacarpiene IV şi V. Modificările cardiace asociate cu malformaţiile osoase de membru superior drept orientează spre un sindrom cord-mână, cel mai probabil un sindrom Holt Oram.Scorul modificărilor scheletale după Gall şi colab. modificat de Gladstone şi Sybert este de 3 din 4, respectiv de 2 din 4 pentru anomaliile cardiace, încadrând cazul nostru într-o formă moderată de boală.
Concluzii
Malformaţiile de membru superior obligă la investigaţii cardiace suplimentare pentru excluderea unui sindrom cord-mână.
Tabloul clinic este variat, pentru cazul nostru asociind si cheilognatopalatoschizis, malformaţie mai puţin raportată în literatura de specialitate.
Depistarea mutaţiei genei TBX5 ar certifica diagnosticul de sindrom Holt – Oram în contextul sindroamelor cord-mână.
Cuvinte cheie - sindrom Holt Oram

LATE DETECTION OF A HOLT-ORAM SYNDROME - CASE REPORT

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Vasile Goldiş Western University of Arad

Aim - to present a case belatedly discovered at the age of 7 years during hospitalization for an exacerbation of asthma.
Case presentation - P.G. , male, 7 years old, is presented for the evaluation of an medium form episode of bronchial obstruction. Clinical examination detects a stature and weight deficient child, scar of the palate post cleft lip and palate surgery, dextroconvex thoracic scoliosis, rhythmic heart beat , SS grade IV / 6, right-hand ectrodactylia with absence of finger I, pectus excavatum.The EKG route is unaltered, and Doppler-Colour echocardiography highlights small atrial septal defect, mitral valve prolapse with mild mitral insufficiency. Radiologically is detected a base metacarpal IV and V syndactyly. The cardiac changes associated with bone malformations of upper limb orientates towards a heart-hand syndrome, most likely Holt-Oram syndrome. Skeletal changes score after Gall and colab. modified by Gladstone and Sybert is 3 out of 4 ,or 2 out of 4 for the cardiac abnormalities, fitting this case in a moderate form of the disease.
Conclusions - Malformations of upper limb requires additional investigations of the heart to rule out heart-hand syndrome.
The clinical picture is varied, for our case associating also cleft lip and palate malformation less reported in the literature.
TBX5 gene mutation detection would prove the diagnosis of Holt-Oram syndrome in the context of heart-hand syndromes.
Key words-Holt Oram Syndrome

90. SINDROMUL OPITZ G/BBB : ASPECTE DIAGNOSTICE

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Sindromul Opitz G/BBB este o afectiune genetica ce cauzeaza o serie de anomalii de-alungul liniei mediene a corpului. Exista doua forme ale sindromului G/BBB: X-linkata si cu transmitere autosomal dominanta. Cele doua forme se deosebesc prin cauzele genetice si modul de transmitere dar semnele si simptomele sunt de obicei aceleasi : hipertelorism, hipospadias, intarziere in dezvoltare s.a. . Acest sindrom face parte din grupul de afectiuni denumit sindromul de deletie 22q11.2 si este cauzat de mutatii ale genelor MID1, SPECC1L, insa, de multe ori nu poate fi identificata o mutatie la acest nivel.
Copil de sex masculin in varsta de 2 ani si 10 luni, fara antecedente heredocolaterale de boli genetice se prezinta: stenoza pulmonara valvulara, defect septal atrial,suflu sistolic gradul III-IV in toate focarele, hipospadias, criptorhidie, hidrocefalie, atrezie de corp calos, hipotrofie staturo-ponderala, intarziere neuro-motorie, hipertelorism, strabism convergent bilateral. Testele genetice pentru confirmarea sindromului Opitz G/BBB sunt in lucru .
Sindromul Opitz G/BBB afecteaza 1:10.000 pana la 1:50.000 nou-nascuti de sex masculin, dar se banuieste ca aceasta afectiune este subdiagnosticata. Datorita manifestarilor polimorfe ale acestei afectiuni, pentru copii cu caracteristici clinice relevante pentru sindromul Opitz G/BBB si mai ales la cei cu atrezie de corp calos, este necesara o analiza genetica extinsa si investigatii clinice, paraclinice si imagistice amanuntite.

OPITZ G/BBB SYNDROME : DIAGNOSTICS STANDPOINTS

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Opitz syndrome is a genetic disorder which causes a number of abnormalities across the midline of the body. There are 2 forms of Opitz syndrome : X-linked and with an autosomal dominant transmission pattern. These 2 entities are caused by different mutations and have a different transmission pattern but they share the same traits and symptoms : hypertelorism, hypospadias, developmental delay a.o. This syndrome is part of a group of disorders known as the 22q11.2 deletion syndrome which is caused by a mutation in the MID1, SPECC1L genes, but many times there can not be identified a mutation at this level.

Male, 2 years and 10 months old, without any family history of genetic disorders presents with : pulmonary valve stenosis, systolic murmur grade III-IV in all auscultatory areas, hypospadias, cryptorchidy, hydrocephalus, corpus callosum agenesis, growth delay, neuro-motor delay, hypertelorsim, bilateral convergent strabismus. Genetics lab tests are being processed.

Opitz syndrome affects 1:10000 to 1:50000 male new-borns, but it's suspected that this disorder is underdiagnosed. Due it's various manifestations extended genetic analysis, clinical, paraclinical investigations and imaging are needed for children that present relevant clinical characteristics of this syndrome, especially those with corpus callosum agenesis.

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91. ASOCIERE RARĂ DE 2 BOLI GENETICE LA ACELAȘI PACIENT: GALACTOZEMIE ȘI AMIOTROFIE SPINALĂ TIP I

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Prezentăm cazul unui pacient ce se prezintă cu sepsis neonatal, cu debut tardiv, cauzat de Escheria Coli, cu multiple insuficiențe de organ asociate: insuficiență hepatică severă, insuficiență renală, colestază importantă, afectare meningeală, fără a fi prezentă hipoglicemia. În condițiile unui sepsis cu E coli, la un nou-născut provenit dintr-o sarcină fiziologică, naștere la termen, necomplicată, fără factori favorizanți pentru sepsis, alimentat la sân, s-a luat în discuție galactozemia. S-a confirmat prin testare enzimatică și ulterior test genetic, care decelează o mutație nedescrisă anterior în galactozemie (c.947_954del la nivelul exonului 10) la acest pacient. Evoluția a fost favorabilă, sub restricție alimentară de lactoză și galactoză, persistând însă o ușoară hipotonie. A fost interpretată inițial ca secundară afectării cerebrale din cadrul sepsisului, deși CT cranian efectuat ulterior nu a relevat modificări. Hipotonia s-a accentuat cu vârsta, este predominant axială, pacientul prezentând și 2 infecții respiratorii inferioare a căror evoluție a fost trenantă. Aspectul clinic sugestiv a impus luarea în discuție și a unei posibile amiottofii spinale asociate. Aceasta s-a confirmat prin testare genetică, evidențiindu-se delețiile specidice la nivelul exonilor 7 și 8 a genei SMN_1. Din păcate, din acest moment prognosticul a fost dictat de această a 2-a patologie, pacientul necesitând traheostomie, bine tolerată, însă evoluția a fost nefavorabilă, decesul survenind în jurul vârstei de 3 ani, secundar unei bronhopneumonii. Asocierea este inedită și atrage atenția asupra algoritmului diagnostic în cazuri complicate, ce ies din tiparul "obișnuit" al bolii diagnosticate inițial.

RARE ASSOCIATION BETWEEN TWO GENETIC CONDITIONS IN THE SAME PATIENT: GALACTOSEMIA AND SPINAL MUSCULAR ATROPHY

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We present the case of a patient presenting with neonatal sepsis, late-onset, caused by Escherichia coli, associated with multiple organ failure: severe hepatic and kidney failure, important cholestasis, meningeal involvement, without hypoglycemia. Given an E. coli sepsis in a newborn from a physiological pregnancy, birth at term, uncomplicated delivery, without predisposing factors for sepsis, breast fed, was suspected. It was confirmed by enzyme testing and subsequent genetic test that detects a mutation previously indescribable in galactosemia (c.947_954del exon 10). Evolution was favorable under food restriction of lactose and galactose, but a slight weakness persisted. It was initially interpreted as secondary to the brain damage in the sepsis, although subsequently performed cranial CT revealed no changes. Hypotonia worsened with age, is predominant axial and the patient presented two lower respiratory tract infections with poor response to treatment. The clinical suggestive aspect imposed the consideration of possible spinal muscular atrophy associated. This was confirmed by genetic testing, highlighting specific deletions at exon 7 and 8 of the SMN1 gene. Unfortunately, this time the outcome was dictated by the 2nd pathology, patient requiring tracheostomy, well tolerated, but the evolution was unfavorable, death occurring around the age of 3 years, secondary to bronchopneumonia. The combination is unique and draws attention to the diagnostic algorithm in complicated cases that get out of "regular" line of the initially diagnosed disease.

92. DIAGNOSTIC DIFERENTIAL IN DISTROFIA MUSCULARA DUCHENNE: PREZENTARE DE CAZ

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Limb girdle muscular dystrophy 2A (LGMD 2A) este produsa de mutatii ale genei pentru calpaina, CAPN 3, iar tabloul clinic nu poate fi, uneori, diferentiat de cel al Distrofiei Musculare Duchenne. (DMD).
In lucrarea de fata prezentam cazul unui baiat de 14 ani, pentru care diagnosticul a fost stabilit cu o intarziere de 8 ani de la debutul bolii. Suspiciunea de boala musculara a aparut la 6 ani datorita manifestarilor clinice: deficit muscular, hipertrofie de molecti, mers pe varfuri, retractii tendinoase precoce.
Diagnosticul initial de DMD s-a bazat pe tabloul clinic si pe valorile crescute de cretinfosfokinaza.
Analiza histopatologica a biopsiei musculare, realizata la varsta de 8 ani, evidentiaza modificari atipice suggestive pentru o histiocitoza sau o afectare neurogena.
Rezultatul examenului MRI practicat la 13 ani demonstreaza o degenerare musculara a regiunii posterioare a coapsei, sugerand diagnosticul de LGMD 2A si impunand practicarea unei alte biopsii musculare. Analiza imunohistochimica practicata e normala pentru proteinele studiate, dar analiza Western Blott evidentiaza o valoare borderline pentru calpaina.
Analiza moleculara practicata ca urmare a acestor rezultate, va identifica un status de heterozigot compus pentru doua mutatii ale genei CAPN 3: c. 380 – 1 G > A (exonul 3) and c. 598 – 612 Del (exonul 4).
Din cauza similitudii tabloului clinic si a limitelor tehnicilor de diagnostic, inaintea utilizarii analizei moleculare si a examenului imunohistochimic, LGMD 2A era, adesea, diagnosticata ca DMD.
Identificarea mutatiilor genei CAPN 3 este, deci, o etapa obligatorie atat pentru stabilirea protocolului terapeutic, cat si pentru un sfat genetic judicios.

DIFFERENTIAL DIAGNOSIS IN DUCHENNE MUSCULAR DYSTROPHY: CASE REPORT

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Limb girdle muscular dystrophy 2A (LGMD 2A) is the most common form of limb muscular dystrophies caused by mutations in the calpain-3 gene (CAPN-3). The pattern of muscle weakness can be clinically indistinguishable from that of Duchenne Muscular Dystrophy (DMD).
We report a case of a 14-year-old boy in which the diagnosis was established with a 8-years delay. The suspicion of muscle disease appeared at 6 years old, revealed by the following signs: muscle weakness, hypertrophic calves, toe walking, early primary contractures and moderate hyperlordosis.
The initial diagnosis of DMD was based on clinical features and very elevated serum creatine kinase levels. A muscle biopsy at the age of 8 showed atypical features which suggested a histiocytosis or neural damage.
An MRI conducted 5 years later revealed fatty degeneration predominantly in the posterior region of the thigh and implied the diagnosis of LGMD 2A as well as the necessity to repeat the biopsy. Immunohistochemical analysis was normal, but the Western Blott showed a normal/borderline amount of calpain-3 in the muscle.
Despite these findings, a molecular analysis implied the fact that he was a compound heterozygote for two mutations of the calpain 3 (CAPN 3) gene: c. 380 - 1 G > A (exon 3) and c. 598 – 612 Del (exon 4).
Before DNA and dystrophin analysis, LGMD was often misdiagnosed as DMD due to the similarities in clinical manifestations and technique limitations.
The identification of CAPN3 gene mutations is an important tool for establishing a genetic advice and therapeutic protocol.

93. CAZ FAMILIAL DE DISPLAZIE FRONTONAZALA NONSINDROMICA

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Displazia frontonazala este o boala genetica foarte rara, cu un tablou clinic variabil, caracterizat, in principal, de un dismorfism cranio-facial ce cuprinde anomalii oculare si malformatii ale piramidei nazale si malformatii ale encefalului: encefalocel si/sau agenezie de corp calos. Displazia frontonazala poate fi sindromica, atunci cand asociaza malformatii cardiace, renale, ale membrelor (absenta tibiei), retard mental si malformatii cerebrale sau nonsindromica. Cele mai multe cazuri apar sporadic, dar au fost descrise si cateva cazuri familiale, cu transmitere autozomal dominantă, justificandu-se, astfel, necesitatea sfatului genetic.

Lucrarea de fata prezinta cazul unei familii in care atat cazul index, reprezentat de un copil de sex feminin in varsta de 4 luni, copil al unui cuplu neconsangvin, cat si mama acestuia, prezinta un dismorfism cranio-facial cu brahicefalie, hipertelorism, fante palpebrale oblice, piramida nazala latita cu fisura mediana prezenta, cheiloschizis.

Dezvoltarea psihomotorie este normala pentru etapa de varsta a copilului; mama prezinta un intelect normal, dar este diagnosticata cu epilepsie. Examenul obiectiv al celorlalte aparate și sisteme nu semnaleaza afectarea acestora.

Explorarile imagistice (echografia transfontanelara si CT craniu), efectuate nu au semnalat anomalii ale cortexului cerebral, iar analiza citogenetica practicata atat pentru cazul index cat si pentru mama, nu a evidentiat nici o aberatie cromozomiala.

Lipsa malformatiilor altor aparate si sisteme claseza cazul prezentat in categoria displaziei frontonazale nonsindromice iar existenta in cadrul aceleiasi familii a doua fenotipuri similare, la mama si copil, sustine transmiterea autozomal dominanta si impune identificarea, prin analiza moleculara, a mutatiei responsabile de fenotipul familial.

NONSYNDROMIC FRONTONASAL DYSPLASIA – A FAMILIAL CASE

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Frontonasal dysplasia is a very rare genetic disease, with variable clinical features, characterized mainly by a cranio-facial dymorphism with ocular abnormalities and nasal pyramid malformation, as well as malformations of the encephalon: encephalocel and/or corpus callosum agenesis. Frontonasal dysplasia may be syndromic, when associated with cardiac or renal malformations, absence of the tibia, mental retardation and cerebral malformations, or nonsyndromic. The disorder is sporadic in most cases, but few familial cases have been reported presenting autosomal dominant inheritance, thus justifying the necessity of the genetic advice.

We report a family case with an index case of a female child of 4 months old from a non consanguineous couple, who, as well as her mother, presents a craniofacial dymorphism with brahycephaly, hypertelorism, oblique eyelid slot, wide nasal pyramid with median split, cheiloschizis. The index case exhibits a normal psychomotor development for her age whilist the mother is having a normal intellect, but is diagnosed with epilepsy. Clinical examination of other body systems and organs showed no other signs of disease. Imagistic exams (transfontanelar echography and cranial computer tomography) didn’t reveal any abnormalities of cerebral, while the cytogenetic analysis showed no chromosomal aberrations for the index case or for the mother. The lack of other malformations makes this case a nonsyndromic frontonasal dysplasia, the existence of two similar phenotypes in the same family, for the mother and the child sustaining the autosomal dominant inheritance pattern and impose the identification through molecular analysis of the mutation responsible for the familial phenotype.

REUMATOLOGIE / RHEUMATOLOGY

94. COMPLICATII TROMBOTICE IN DOUA CAZURI DE LUPUS ERITEMATOS SISTEMIC: PARTICULARITATI DE DIAGNOSTIC

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Trombozele venoase reprezinta o patologie din ce in ce mai frecventa in populatia pediatrica, cel mai probabil datorita perfectionarii mijloacelor de diagnostic. Trombembolismul pulmonar(TEP) in populatia pediatrica este rar (incidenta anuala de 0,9 la 100 000 copii) , dar studiile sugereaza ca de cele mai multe ori este nerecunoscut sau/si nu este luat in considerare ca prim diagnostic de catre practician. Printre factorii de risc ai TEP se numara si lupusul eritematos sistemic (LES) in asociere

cu sindromul antifosfolipidic (SAP). Diagnosticul prompt si tratamentul specific in TEP sunt cruciale, intarzierea diagnosticului avand consecinte grave, chiar letale.
Scopul acestei lucrari este de a atrage atentia asupra riscului de TEP la copil. Am luat in considerare doua cazuri de lupus eritematos sistemic (LES). Primul caz este al unui pacient cunoscut de 2 ani cu LES, monitorizat periodic si aflat sub tratament imunosupresiv,ce a dezvoltat simptomatologie sugestiva de TEP cu initierea prompta a tratamentului specific si evolutie favorabila. Cel de al doilea caz , un pacient cunoscut cu trombocitopenie , ce dezvolta sindrom antifosfolipidic complicat cu TEP masiv, conturand in evolutie si criteriile de LES, si care, in pofida tratamentului prezinta o evolutie nefasta.
Concluzii : TEP se regaseste in populatia pediatrica. Chiar daca factorii de risc si simptomatologia clinica sunt cunoscute ,aceasta patologie ramane o provocare pentru medicul practician. Lucrarea de fata evidentiaza factorii de risc si simptomatologia clinica ce ar trebui sa ridice suspiciunea de TEP.

THROMBOTIC COMPLICATIONS IN TWO CASES OF SYSTEMIC LUPUS ERYTHEMATOSUS: DIAGNOSTIC FEATURES

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Venous thrombosis is becoming more common in the pediatric population, most likely due to the improvement of diagnostic tools. Pulmonary thrombembolism (PTE) is uncommon in the pediatric population, (annual incidence 0,9 to 100 000 children), but studies suggest that PTE , most often is not recongnized or considered by the practitioner. Systemic lupus erithematosus (SLE) is a risk factor, especially in association with antiphospholipidic syndrome. Clinical evaluation, diagnosis and treatment of PTE are imperative, because delaying diagnosis has serious consequences , even death.
The purpose of this paper is to aware the clinician about the risk of pulmonary thrombembolism in children. We present two cases of SLE. The first is an a pacient known for 2 years with SLE, periodically monitored and treated with immunosuppressives, who developed clinical symptoms highly suggestive for PTE. The treatment was immediately initiated and the evolution was favorable. The second case, an adolescent known with thromocytopenia , who develops antiphospholipidic syndrome complicated with massive pulmonary thrombembolism with unfavorable outcome, in spite of all our efforts. This pacient also developed criteria for systemic lupus erythematosus.
Conclusions: Pulmonary thrombembolism can be found in the pediatric population. Although the risk factors and clinical symptoms are known , this pathology remains a challenge for the practitioner . This paper highlights the risk factors and clinical symptoms that should raise the suspicion of pulmonary thrombembolism.

95. TRATAMENTUL BIOLOGIC IN ARTRITA IDIOPATICA JUVENILA. OBSERVATII PE MARGINEA A PATRU CAZURI CLINICE.

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Autorii prezinta rezultatele tratamentului combinat: Metotrexat si "substantele biologice"- proteine produse in laborator, cu efect reglator al procesului inflamator de la nivelul tesuturilor si organelor. Substantele biologice modifica raspunsul biologic al citokinelor (TNF-α, IL-6, IL-1, oxidul nitric, etc.) si diminua distructia articulara si alterarea capacitatii functionale.
Autorii raporteaza trei cazuri tratate cu Etanercept (actiune anti TNF-α) in doza de 0,85 mg kgcorp, subcutan, bisaptamanal. Raspunsul terapeutic a fost bun, cu remisie clinica si biologica prelungita. Al patrulea caz a fost tratat cu Tocilizumab, efect anti IL-6, doza 8 mg/kg/doza, bilunar in perfuzii intravenoase, timp de un an. Raspunsul terapeutic a fost pozitiv, dar remisia a fost doar de un an, dupa care a aparut o recadere clinica si biologica sub forma de artrita idiopatica juvenila, forma sistemica dominata de febra de "tip colagenic". Se subliniaza efectul benefic al terapiei biologice in artrita idiopatica juvenila, forme care nu raspund la terapia combinata cu doua sau mai multe medicamente.
Cuvinte cheie: artrita idiopatica juvenila, tratamentul biologic, anticorpi anti TNF-α

96. POLIMIOZITA PRIMARA IDIOPATICA. PREZENTARE DE CAZ

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Introducere. Polimiozita este o afectiune musculara inflamatorie, cu caracter autoimun.
Scopul: In lucrare se prezinta cazul unui adolescent cu miopatie cu debut la varsta de 15 ani.
Prezentare de caz: Pacient in varsta de 15 ani, sex M, se interneaza pentru dureri musculare severe la nivelul bratelor si gambelor. Examen clinic: tegumente calde, hipertonie musculara, limitarea miscarii de flexie a gambelor pe coapse, limitarea extensiei bratelor si limitarea ridicarii bratelor in pozitie verticala. Paraclinic: CK = 603 U/L, LDH = 999 U/L, VSH = 30

mm/h; CRP = 46.2 mg%. Examenul neurologic: miopatie inflamatorie secundara. EMG : traseu cu potentiale hipervoltate de scurta durata si un pattern de recrutare crescut, sugestiv pentru o miopatie. Biopsia musculara releva: zone de atrofie musculara, infiltrat limfocitar endomisial si perivascular. Anticorpii specifici miozitei efectuati (Ac. Anti-Mi-2, Ac. Anti Jo-1, Ac. Anti SRP, Ac. Anti Pl-7, Ac. Anti Pl-12, Ac. Anti EJ, Ac. Anti OJ) si cei asociati miozitei (Ac. Anti Ku, Ac. Anti Pm-Scl 100, c. Anti Pm-Scl 75, Ac. Anti Ro-52) au fost negativi. Rezultatele investigatiilor paraclinice si contextul au orientat diagnosticul spre polimiozita primara idiopatica. A fost initiata puls-terapie cu Metilprednisolon intrvenos (1g/zi timp de 3 zile), si apoi corticoterapie per os cu Prednison (80 mg/zi, cu scaderea progresiva a dozelor). La terminarea curei simptomatologia clinica se remite, enzimele musculare revin la valori normale dar persista sindromul inflamator si traseul EMG de aspect miogen.
Particularitatea cazului: pacient de sex masculin cu anticopri specifici negativi si cu debut clinic de artropatie.
Cuvinte cheie: polimiozita, adolescent, artralgii.

PRIMARY IDIOPATHIC POLYMYOSITIS. CASE REPORT

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Introduction. Polymyositis is an inflammatory muscle disease, with autoimmune character.
Aim: To present the case of an 15 years adolescent with myopathy.
Case report: A 15 years old male patient was admitted to the hospital complaining of severe muscle pain in arms and shins. Clinical exam: warm teguments, muscular hypertonia, limitation of doing the flexion of his shin on his thighs, limitation of doing the extension of his arms and rising the arms over his head. Laboratory findings: CK = 603 U/L, LDH = 999 U/L, VSH = 30 mm/h; CRP = 46.2 mg%. The neurological examination: secondary inflammatory myopathy. EMG: hyper-voltage potentials with short duration, increased pattern of recruitment, suggestive for a myopathy. Muscle biopsy reveals: local atrophy of muscle fibre, endomysial inflammatory limfocitar infiltration and perivascular infiltration. Myositis-specific antibodies (Ac. Anti-Mi-2, Ac. Anti Jo-1, Ac. Anti SRP, Ac. Anti Pl-7, Ac. Anti Pl-12, Ac. Anti EJ, Ac. Anti OJ) and myositis-associated antibodies (Ac. Anti Ku, Ac. Anti Pm-Scl 100, c. Anti Pm-Scl 75, Ac. Anti Ro-52) were all negative. The results of all the investigation that have been made were integrated in clinical context and the diagnostic of Primary idiopathic polymyositis was established. Pulse corticosteroid therapy with intravenous Methylprednisolone was initiated (1 g per day for 3 days), and then corticosteroid therapy per os with Prednisone (80 mg per day, decreasing the dose progressively). At the end of the cure, the symptomatology was remitted, the muscular enzymes went back to normal values but the inflammatory syndrome persists and so does the EMG myogenic route.
Particularity of the case: male patient with negative specific antibodies and arthropathic onset.
Keys words: polymyositis, adolescent, arthralgia.

URGENŢE PEDIATRICE – TOXICOLOGIE CLINICĂ / PEDIATRIC EMERGENCIAS – CLINIC TOXICOLOGY

97. CORP STRAIN VEGETAL INTRABRONSIC LA UN SUGAR. PREZENTARE DE CAZ.

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Aspiratia de corp strain este posibila la orice varsta. De catre sugari pot fi aspirate fragmente de jucarii sau componente ale dietei. Întotdeauna reprezinta o urgenta majora, salvarea pacientului fiind prin broho-aspiratia efectuata de serviciul ORL.
Autorii prezinta pacienta MC, in varsta de 9 luni la internarea in Clinica I Pediatrie, unde a fost tratată intre 30-IX-25-X-2010. Din anamneza s-au retinut: este al 4-lea copil, cu trei surori si parinti sanatosi, diversificata incorect la 2,5 luni si cu o dieta inadecvata, ce includea seminte. La internare, s-au notat urmatoarele: sugar de sex femeiesc, cu G= 8600 g, subfebrila 37,3, cu stare generala influentata, cu un examen clinic al aparatului respirator ca in bronsiolita, cu semne de rahitism cranio-toracic. La radiografiile pulmonare s-au descris: opacifierea difuza neomogena a lobului mediu pulmonar drept, mediastin si trahee tractionate spre dreapta. Pe baza anamnezei, a evolutiei clinice trenante si a radiografiilor sugestive, s-a suspicionat

corp strain intrabronsic si s-a efectuat interventia ORL. Au fost necesare doua bronho-aspiratii pentru a se extrage toate fragmentele de seminte de floarea-soarelui, dar sugarul s-a externat vindecat.
Concluzie: Anamneza trebuie sa fie amanuntita si insistenta, iar bronho-aspiratia realizata la timp, deoarece corpii straini vegetali produc suprainfectii, urmate de supuratii bronho-pulmonare; alteori se pot mobiliza in trahee producand moartea subita a pacientului. Cazul descris a fost cu final fericit.

VEGETAL INTRABRONCHIAL FOREIGN BODY IN AN INFANT. CASE REPORT.

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Foreign body aspiration is possible at any age. Infants can aspirate fragments of toys or organic materials. It always represents a major emergency, the life-saving maneuver of patient being bronchoaspiration in Otolaryngology services.
Authors present M.C. female-infant, aged 9 months at hospitalization in Pediatrics Clinic, where she was treated from 30-IX- to 25-X-2010. From history were retained: she was the fourth daughter of healthy parents, whose three sisters had no medical history as well, had inadequate dietary diversification at 2.5 months, which included seeds.
On admission, we noted the following: female infant with W = 8600 g, t=37.3°C , influenced general state, at physical examination the patient presenting the clinical signs and symptoms of bronchiolitis and with signs of cranial and thorax rickets. The chest radiographs revealed: diffuse, mild heterogeneity in the right medium lobe, mediastinum and trachea pulled to right. Based on history, difficult clinical evolution and suggestive chest-radiographs: intrabronchial foreign body was suspected and ENT- surgery was performed. It was necessary two bronchial aspirations to remove all fragments of sunflower seeds, but the infant was discharged cured.
Conclusion: The medical history must be thorough and pushy, and bronchial aspiration completed on time, because foreign vegetal bodies might produce superinfection, followed by bronchopulmonary suppurations; sometimes it can be moved into trachea causing sudden death of the patient. The case described had a happy-end.
Key-words: foreign body, infant, bronchial aspiration

98. INTOXICATIE ACUTA SEVERA CU AMITRAZ LA UN ADOLESCENT

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Obiective: Amitrazul este un pesticid - triazapentadiene, un alfa 2 adrenergic agonist din familia amidinelor, compus chimic folosit pentru distrugerea ectoparazitilor la animale. Dupa expunerea la Amitraz, se produce depresia sistemului nervos central si apar manifestari cardiovasculare si respiratorii.
Prezentarea cazului: Adolescent in varsta de 17 ani, se interneaza in clinica noastra pentru coma, midriaza, bradicardie, respiratie superficiala si leziuni corneene, dupa ingestia de Amitraz si medicamente (amoxicilina, cefuroxim si ketoprofen) cu 12 ore anterior, in scop suicid. A fost gasit de catre parinti, comatos. Manifestarile clinice la internare: GCS=6, midriaza fixa, AV=55b/min, TA=65/35 mmHg, FR=12/min, leziuni post autoagresiune la nivelul antebratului stang, diureza prezenta. Tratament: suportiv – ventilatie mecanica 7 ore, perfuzie endovenoasa.
Evolutie: in primele 7 ore de la internare AV a variat intre 43 si 60 b/ min, TA s-a mentinut scazuta (65/9mmHg – 70/40mmHg) apoi a revenit la normal 90/55mmHg; dupa 7 ore coma s-a superficializat si dupa inca 2 ore pacientul a devenit constient.
Examene de laborator: hemograma normala, pH-ul sangvin intre 7,34 – 7,36. Glicemia a fost in limite normale. Examenul toxicologic de urina a evidentiat Amitraz si metaboliti. Nu s-au inregistrat modificari EKG.
Dupa 19 ore de la ingestie, pacientul a devenit constient, pupilele au devenit normale ca dimensiuni si reactivitate, AV si TA s-au normalizat. Examenul neurologic a fost in limite normale. Pacientul a fost transferat la psihiatrie.
Concluzii: nu exista antidot specific pentru intoxicatia cu amitraz. Tratamentul este simptomatic si suportiv si revenirea la starea de sanatate este completa. Folosirea necorespunzatoare a amitrazului in scop suicid subliniaza necesitatea toxicovigilentei.
Cuvinte cheie: adolescent, intoxicatie voluntara, amitraz, tratament suportiv.

SEVERE ACUTE AMITRAZ POISONING IN A TEENAGER

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Objective: Amitraz is a pesticide - triazapentadiene, an alfa2 adrenergic agonist in the amidine chemical family used for controlling ectoparasites in animals. After exposure it may produce central nervous system depression and also cardiovascular and respiratory symptoms.

Case report: Male teenager, 17 years old, admitted in our hospital for coma, midriasis, bradicardia, superficial respiration and corneal lesions after ingestion of Amitraz and drugs (amoxicillin, cefuroxim and ketoprofen) 12 hours before, as suicidal attempt. Was found in coma in his room. Clinical features at admission: GCS=6, nonreactive midriasis, HR=55b/min, BP=65/35 mmHg, respiration=12/min, auto inflicted lesions on the left forearm, diuresis present. Was admitted in ICU. Treatment: supportive – mechanical ventilation for 7 hours, intravenous fluids.
Evolution: the first 7 hours of evolution heart rate values varied between 43 – 60 beats/min, blood pressure values kept low (65/9mmHg–70/40mmHg) then returned to normal 90/55mmHg; after 7 hours coma started to become superficial then after another 2 hours the patient regained consciousness.
Lab exams: normal blood count, blood pH values 7,34 – 7,36. Blood glucose levels kept normal. Urine toxicological exam revealed Amitraz and metabolites. There were no ECG alterations.
19 hours after voluntary ingestion of Amitraz our patient regained consciousness, pupils dimension and reactivity became normal, heart rate and blood pressure returned to normal values. Neurological exam was normal. Patient was referred to the psychiatric ward.
Conclusions: there is no specific antidote for Amitraz poisoning. Treatment was only symptomatic and supportive and the patient successfully recovered to full health status. The misuse of amitraz for deliberate self-harm emphasizes the necessity for continued toxicovigilance.
Key words: teenager, self-deliberate exposure, amitraz, supportive treatment.

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99. SURPRIZA DE DIAGNOSTIC LA UN PACIENT COMATOS

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Societatea contemporana trateaza cu superficialitate problema sinuciderii. Romania in prezent se confrunta cu un val intens mediatizat al conduitelor suicidare.
Toate tarile, indiferent de continent, atentioneaza ca ideatia suicidara este comuna la adolescenti iar experimentarea uneia sau mai multor tentative de suicid in lungul existentei acestora este o problema greu de cuantificat de la o tara la alta. Din estimarile OMS pentru Europa, rata anuala medie pentru tarile europene este de 20,5 la 100.000 de locuitori.
Tentativele de suicid par a fi mai putin comune inainte de pubertate. Metoda aleasa pentru gestul suicidar determina printre altele si daca sinuciderea va ramane la stadiul de tentativa sau va fi desavarsita. Femeile sunt tentate sa foloseasca o metoda mai putin traumatizanta, precum ingerarea excesiva de medicamente.
Procesul cresterii si dezvoltarii copilului si adolescentului, trebuie privit ca factor de vulnerabilitate crescuta, fiind in acelasi timp indispensabil normalitatii. Mediul, ca o conditie a actului suicidar, este corelat cu stresul. Exista un control mai putin eficient al tinerilor asupra situatiilor de stres, ei apeland in acest sens la resursele individuale, la care se adauga cele invatate, completate de factorii protectori individuali. Presiunea in cazul acestora este corelata cu expectatiile familiei, cu valorile microgrupului in care se formeaza si exista ca persoana.
Autorii prezinta cazul unei fetite de 12 ani, diagnosticata cu “Sindromul Italia”, aflata la a doua tentativa de suicid.
Cuvine cheie: copil, suicid, coma

SURPRISE DIAGNOSIS ON A COMATOSE PATIENT

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Contemporary society treats lightly the problem of suicide. Romania is currently experiencing a wave of highly publicized suicidal behaviors.
All countries around the world, warns that suicidal ideation is common in adolescents and experiencing one or more suicide attempts throughout their existence is a problem difficult to quantify from one country to another. OMS estimates from Europe, the average annual rate for European countries is 20.5 per 100,000.
Suicidal seem to be less common before puberty. The method chosen for suicidal gesture determine among other things if the suicide will remain only in the attempt stage or it will end in a real suicide . Women are tempted to use a less traumatic method, such as excessive intake of drugs.
Child growth and development should be regarded as a factor of increased vulnerability, while being indispensable normal. The environment, as a condition of the suicidal act is correlated with stress. There is a less effective control over stressful situations in young people’s case, falling back on in this case to personal resources, plus the learned abilities, complemented

by individual protective factors. The pressure in such case is related to the family expectations, micro group values where they exist as a person.
The authors present the case of a 12 year old girl, diagnosed with "Italy Syndrome", located at the second attempt suicide.
Key words: child, suicide, coma

NUTRIŢIE ŞI BOLI METABOLICE / NUTRITION AND METABOLIC DISEASES

100. STRESSUL OXIDATIV SI TERAPIA ANTIOXIDANTA IN FIBROZA CHISTICA LA COPIL

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Fibroza chistica (FC) este cauzata de un defect al genei CFTR. Prezenta unei gene defective CFTR produce un dezechilibru redox in celulele epiteliale si lichidele extracelulare si cauzeaza generarea anormala a speciilor oxigen reactive (SOR). In bronsiile bolnavilor cu FC, lichidul din caile respiratorii se caracterizeaza prin concentratie crescuta a SOR, nivele scazute ale glutationului si acid nitric scazut. Cresterea speciilor pro-oxidative in acest lichid contribuie la distructia progresiva a tesutului pulmonar si la amplificarea raspunsului inflamator in caile respiratorii in FC. Stressul oxidativ si inflamatia pot afecta activitatea biofizica a surfactantului, conducand la alterarea precoce a functiei pulmonare si afectarea atat a componentelor lipidice, cat si proteice. Proteina D din surfactant devine incapabila sa aglutineze bacteriile cand este modificata prin oxidare, facilitand colonizarea patogenilor in plaman. Suplimentele cu antioxidanti pot reduce distructia oxidativa in plamani. Diferitii antioxidanti ca vitamina E, β-carotenul si acizii grasi ω-3 atenueaza semnele biochimice de stress oxidativ. Glutationul – scutul antioxidant major la nivel epitelial, nu este eliberat corespunzator in plamanii bolnavilor cu FC. Administrarea glutationului amelioreaza functia pulmonara in unele cazuri si scade stressul oxidativ. Unele enzime care intervin ca antioxidanti sunt dependente de seleniu; astfel, suplimentele cu seleniu ajuta la stimularea actiunii antioxidante. Vitamina C hidrosolubila scade odata cu varsta la bolnavii cu FC. Prin urmare, suplimentele cu vitamina C ajuta la cresterea acestor nivele. Concluzie: impreuna cu tratamentul clasic, suplimentarea cu antioxidanti poate fi benefica pentru incetinirea ratei deteriorarii functiei pulmonare.
Cuvinte cheie: fibroza chistica, copil, stress oxidativ, antioxidanti.

OXIDATIVE STRESS AND ANTIOXIDANT THERAPY IN CYSTIC FIBROSIS IN CHILDREN

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Cystic fibrosis (CF) is caused by a defect of CFTR gene. The presence of defective CFTR appears to produce a redox imbalance in epithelial cells and extracellular fluids and causes an abnormal generation of reactive oxygen species (ROS). Airway surface liquid (ASL) in CF bronchi is characterized by increased concentration of ROS, lowered levels of glutathione and reduced nitric oxide. The increase of pro-oxidative species in ASL contributes to the progressive lung tissue damage and to the amplification of the inflammatory response in CF airways. Oxidative stress and inflammation can affect surfactant biophysical activity leading to early alterations of lung function and involving both lipid and protein components. Surfactant protein D becomes unable to agglutinate bacteria when it is modified by oxidation, which facilitates pathogen colonization in the lung. Antioxidant supplements might reduce the oxidative damage in the lungs. Different antioxidants as vitamin E, β-carotene and ω-3 fatty acids have observed to alleviate selected biochemical signs of oxidative stress. Glutathione - the major antioxidant shield in the epithelial lining fluid, in people with CF is not released into the lungs properly. Administration of glutathione improve lung function in some cases and lower oxidative stress. Some enzymes which help antioxidants work are dependent on selenium; so selenium supplements aim to stimulate antioxidant action. Vitamin C decreases with age in people with cystic fibrosis, so vitamin C supplements aim to rebuild these levels.Conclusion: antioxidant supplementation may prove beneficial in slowing the rate of deterioration in pulmonary function when combined with current therapies.
Key words: cystic fibrosis, child, oxidative stress, antioxidants.

101. CORELATII INTRE ALIMENTATIA MODERNA SI CRESTEREA INCIDENTEI DIABETULUI ZAHARAT LA NIVEL MONDIAL

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In ultimile decenii diabetul zaharat a capatat proportii epidemice, la nivel mondial in 2014 fiind diagnosticate 387 milioane de persoane, iar previziunile pentru 2035 se ridica 592 milioane de cazuri diagnosticate. Intrebarea lumii medicale este : ce anume a dus la aceasta crestere ingrijoratoare? In ultimul deceniu s-au efectuat numeroase studii care ridica diferite ipoteze printre care si alimentatia moderna de tip fast food care a luat avant incepand din anii 1950 in SUA, unde si incidența a crescut de la 6,6 / 100.000 în 1950-1952 la 11,3 în 1959-1961. Exista o relatie directa intre aparitia lanturilor de tip fast food, raspandirea acestora, frecventa consumarii acestui tip de alimente, si cresterea alarmanta a incidentei la nivel mondial a obezitatii si diabetului zaharat , fapt dovedit de studii internationale din ultimul deceniu. In acest context la nivelul jud Galati s-a constatat o crestere alarmanta a cazurilor noi de diabet zaharat, aproape dublu fata de anii 1980, cu un varf in 2010 de 18 cazuri noi. Anamneza si studiile populationale arata o concordanta intre cresterea incidentei diabetului si multitudinea fast food-urilor la nivelul judetului. De mentionat ca aproximativ 33% dintre pacienti prezentau status supraponderal la debut. Deoarece numarul copiilor proveniti din urban este aproximativ egal cu numarul celor proveniti din rural, nu s- a gasit o explicatie concreta privind implicarea produselor tip fast food in cresterea incidentei diabetului.

CORRELATIONS BETWEEN MODERN DIETS AND INCREASEAD INCIDENCE OF DIABETES WORLDWIDE

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In recent decades diabetes gained epidemic proportions,worldwide in 2014 they were 387 million people diagnosed and projections for 2035 amounted to 592 million diagnosed cases. Question that the medical world is concerned about is: what led to this dramatic increase? In the last decade, there have been many studies that raise various assumptions including modern diet of fast food that has soared since the 1950s in the US, where the incidence increased from 6.6 / 100,000 in 1950-1952 to 11.3 in 1959-1961. There is a direct relationship between the occurrence of fast food chains, their spread, frequency of eating this type of food, and the alarming increase in the incidence of global obesity and diabetes, as evidenced by international studies in the last decade. In this context in Galati we found an alarming increase in new cases of diabetes, nearly double the 1980s, with a peak in 2010 of 18 new cases. History and population studies show an increased incidence of diabetes in concordance between fast food and many sites in the county. Mentioned that about 33% of patients have onset overweight status. As the number of children from urban is approximately equal to the number of those coming from rural areas, it has not been found a concrete explanation on the involvement of fast food products in the increasing incidence of diabetes.

102. BOALA HUNTER- O PROVOCARE DE DIAGNOSTIC SI TRATAMENT

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Mucopolizaharidoza tip II-boala Hunter- este o boala monogenica rara, X-linkata caracterizata prin deficitul enzimei lizozomale - iduronat 2 sulfataza- responsabila pentru metabolizarea glicozaminoglicanilor:heparan sulfat, dermatan sulfat , urmata de acumularea patologica a GAG in multiple organe ale organismului. Simptomatologia si progresia bolii este foarte heterogena; pacientii se pot prezenta la diferite medici spacialisti inainte de stabilirea diagnosticului. Substitutia enzimatica si transplantul de celule stem au ajutat in ultimul timp in ameliorarea evolutiei bolii, insa caracterul complex al acestei boli, precum si ireversibilitatea anumitor manifestari necesita colaborarea mai multor specialitati. Autorii prezinta 4 cazuri de MPZtip II care se afla sub tratament pe Sectia Terapie Acuti al Spitalului Clinic de Copii Brasov

HUNTER DESEASE-A DIAGNOSIS AND THREATMENT CHALLENGE

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Mucopolysaccharidosis type II is a rare, life-limiting, X-linked recessive disease characterised by deficiency of the lysosomal enzyme iduronate-2-sulfatase. Consequent accumulation of glycosaminoglycans leads to pathological changes in multiple body systems. Age at onset, signs and symptoms, and disease progression are heterogeneous, and patients may present with many different manifestations to a wide range of specialists. In recent years, disease-specific treatments such as enzyme replacement therapy and stem cell transplantation have helped to address the underlying enzyme deficiency in patients with MPS II. However, the multisystem nature of this disorder and the irreversibility of some manifestations mean

that most patients require substantial medical support from many different specialists, even if they are receiving treatment. We,d like to present 4 cases of MPS type II who are getting threatment in Clinical Hospital of Pediatrics Brasov.

103. EVALUAREA FACTORILOR CARE INFLUENTEAZA VARSTA INTRODUCERII ALIMENTELOR DE DIVERSIFICARE LA SUGARI

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Introducere
Actualmente, exista controverse cu privire la varsta la care se initiaza diversificarea si succesiunea introducerii alimentelor. Impactul diversificarii poate avea consecinte asupra dezvoltarii alergiilor alimentare si a excesului ponderal.
Obiective
Ne-am propus sa evaluam factorii care influenteaza varsta de initiere a diversificarii, tipul de aliment ales pentru diversificare si greselile care se fac in alimentatia sugarilor.
Material si metoda
In intervalul ianuarie 2014-decembrie 2014, am analizat 375 de sugari (2 luni si 12 luni), prezentati in ambulatoriul de specialitate. Parintii au primit un chestionar care a inclus: data nasterii, greutatea la nastere, durata alimentatiei naturale, initierea alimentatiei complementare sau artificiale, tipul de lapte folosit,primul aliment de diversificare si varsta diversificarii cu alte tipuri de alimente. Sugarii au fost evaluati pe grafice de crestere.
Rezultate
Varsta medie de diversificare este de 4 luni, cu extreme intre 2 luni (15 cazuri) si 9luni (43 cazuri). Alimentele preferate pentru diversificare au fost fructele, in 60 % din cazuri, iar combinatiile de tip mar ras cu biscuit in 25%. Greseli alimentare majore: sare sub 12 luni in 34.1%, zahar sub 12 luni in 21%, suc de fructe sub 4 luni in 12%, carne sub 6 luni in 10%, lapte de vaca sub 9 luni in 35% din cazuri.
Concluzii
In 212 cazuri au fost sesizate erori in initierea diversificarii. Nivelul educational scazut al mamelor, asociat deficientelor economice si lipsei de informare, reprezinta unul din elementele de baza ale diversificarii incorecte.
Cuvinte cheie: sugar diversificare greutate aliment

TIMING EVALUATION OF INTRODUCING SOLID FOODS IN INFANTS

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Introduction
Currently, there is controversy about the age at which diversification and the sequence of introducing solid foods should be initiated. The impact of diversification can have consequences over the development of food allergies and overweight.
Objectives
We aimed to evaluate the factors that influence age of introducing complementary food, type of complementary food used and mistakes in infants` diet.
Methods
During January 2014- December 2014, we examined 375 children between 2 months and 12 months present in ambulatory. Parents recived a questionnaire that included: date of birth, birth weight, the term of breastfeeding, initiating complementary or artificial nutrition, type of milk, the first food used in diversification and age of diversification with other types of food. Infants were evaluated on growth charts.
Results
The average age of diversification is 4 months, with extremes between 2 months (15 cases) and 9 months (43 cases). Favorite foods for diversification were fruit in 60% of cases, combination between biscuits and applesauce in 25 %. Major food mistakes: salt below 12 months in 34.1%, sugar below 12 months in 21% , fruit juice below 4 months in 12%, meat below 6 months in 10%, cow's milk below nine months in 35% of cases.
Conclusion
In 212 cases there have been reported errors in initiating diversification. Low educational level of mothers associated with economic deficiencies and lack of information, is one of the basic element of incorrect diversification.
Keywords: infant diversification weight food

104. COPILUL NEGLIJAT – POSIBIL CAZ MEDICO-LEGAL

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Datorita conditiilor socio-economice precare, lipsei de educatie si migrarii populatiei, neglijarea copiilor reprezinta o realitate a ultimilor ani in tara noastra.

Neglijarea este cea mai frecventa forma de maltratare a copilului (aprox 65% din totalul formelor de abuz ale copilului) si poate avea forme multiple: educationala, fizica, medicala, emotionala.

Anchetele speciale elaborate pe baza unor protocoale preexistente, aplicate in cursul spitalizarii, in cazul copiilor considerati cu risc mediu/ridicat de abuz sau neglijenta in familie se pot dovedi extrem de utile pentru prevenirea efectelor nefaste posibile.

Aplicarea chestionarului elaborat la un numar de 320 de cazuri au aratat la 1/5 din ele o forma de abuz, majoritatea (83%) fiind cazuri din mediul rural. Abuzul medical poate avea consecinte catastrofale in plan familial cu „ricosaj" catre medicii implicati in procesul de diagnostic si tratament adecvat.

Drumul de la copilul neglijat la un posibil caz medico-legal poate fi uneori imprevizibil si, deseori, clinicianul, pe baza unei anamneze riguroase si apeland la gandirea si analitica simtului clinic va trebui sa deceleze si sa surmonteze capcanele aparente in desfasurarea actului medical.

Un caz ilustrativ a stat la baza unui studiu prospectiv desfasurat pe parcursul unui an calendaristic ce a aratat un real fenomen de iceberg, referitoare la cel putin o forma de abuz existenta la aprox 20% din cei chestionati.

Sesizarea la timp a riscurilor majore, aplicarea consilierii, implicarea medicului curant, psihologului, serviciilor de ajutor si protectie sociala, vor reusi, in mare parte, stoparea acestui fenomen nociv si din ce in ce mai pregnant.

NEGLECTED CHILD - POSSIBLE FORENSIC CASE

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Due to poor socio-economic conditions, lack of education and population migration, child neglect is a reality in recent years in our country.

Neglect is the most common form of child maltreatment (about 65% of all forms of child abuse) and can have multiple forms: educational, physical, medical, emotional.

Special surveys developed on existing protocols applied during hospitalization in children with medium/high risk of abuse or neglect in the family can be extremely useful to prevent possible adverse effects.

The development of a questionnaire for a number of 320 cases showed that 1/5 of them had a form of abuse, the majority (83%) were cases in rural areas. Medical abuse can have catastrophic consequences in the family with "ricochet" toward clinicians involved in the diagnosis and appropriate treatment.

The path from child neglect to a possible forensic case can sometimes be unpredictable and often the clinician, based on a thorough case history and appealing to the sense of clinical and analytical thinking will need to detect and overcome apparent pitfalls in the development of medical care.

An illustrative case was the basis of a prospective study conducted during a year that showed a real iceberg phenomenon, related to at least one form of abuse that exists at about 20% of those questioned.

Timely notification of the major risks, application counseling, involving the physician, psychologist, support services and social protection, will largely succeed to stop this harmful and increasingly phenomenon.

105. CONSIDERATII CLINICO-BIOLOGICE PRIVIND RAHITISMUL CARENTIAL LA SUGAR SI COPILUL MIC

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Obiective. Stabilirea unor corelatii intre rahitismul carential la copil, patologia asociata, dezvoltarea somato-psihica si influenta conditiilor socio-economice.

Material si metoda. Am efectuat un studiu epidemiologico-clinic retrospectiv privind rahitismul carential la copiii internati in Clinica II Pediatrie, S.C.J.U. Craiova, in 2014.

Rezultate. Prevalenta rahitismului carential in cadrul patologiei generale a fost 19%. Cele mai multe cazuri s-au inregistrat la copilul mic 1-3 ani (64%) si sugarul 8-12 luni (12%), la sexul masculin (61%), fara diferente semnificative pe medii de provenienta. Alimentatia a fost naturala in 41% din cazuri, artificiala in 53% ; diversificare incorecta in 39%. Manifestarile clinice cel mai frecvent intalnite: bose frontale/parietale, craniotabes, plagiocefalie, intarzierea inchiderii fontanelei, torace evazat la baze, sant Harrison, stern infundat. Afectiuni asociate : 64% IACRS, 23% IACRI, 14% afectiuni digestive, 12% cutanate, 4% neurologice. Anemia carentiala/ferirpiva a fost prezenta in 45%, distrofia in 29%. Calcemia serica scazuta s-a inregistrat in 27% din cazuri. Raportat la nivelul educational al parintilor, ponderea cea mai mare a copiilor cu rahitism

carential s-a inregistrat la cei cu studii inferioare si medii; 54% au prezentat nivel socio-economic precar. Profilaxia rahitismului carential a fost efectuata incorect/incomplet in 38%, neefectuata in 19% din cazuri. Durata medie de spitalizare a fost 5-10 zile.
Concluzii. Lipsa suplimentarii cu calciu si vitamina D la mame in ultimul trimestru de sarcina, alaturi de varsta mica a mamei, educatie precara, nivelul socio-economic scazut, nasteri multiple, prematuritate/dismaturitate, carente alimentare cantitative/calitative, alimentatie incorecta, reprezinta factorii de risc intalniti in rahitismul carential.
Cuvinte cheie: rahitism carential, alimentatie, copil

CLINICAL AND BIOLOGICAL CONSIDERATIONS REGARDING CARENTIAL RICKETS IN INFANTS AND TODDLERS

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Objectives. Establishing some correlations among carential rickets in children, associated pathology, somatopsychic development, and the influence of the socio-economic conditions.
Material and method. We performed a retrospective epidemiological-clinical study on carential rickets in children admitted in the 2nd Pediatric Clinic, Emergency County Hospital Craiova, in 2014.
Results. The prevalence of carential rickets within general pathology was 19%. Most cases were registered in toddlers, aged 1-3 years (64%) and infants, aged 8-12 months (12%), in males (61%), without significant differences regarding the origin environment. The feeding was natural in 41% of the cases, artificial in 53%; incorrect diversification in 39%. The most frequent clinical manifestations: frontal/parietal bosses, craniotabes, plagiocephaly, delayed closure of fontanelle, flared thorax to base, Harrison's sulcus, and sunken chest. Associated affections: 64% AURI, 23% ALRI, 14% digestive affections, 12% cutaneous, and 4% neurological. Carential/ferriprive anemia was present in 45%, dystrophy in 29%. Low serum calcium level was registered in 27% of the cases. Regarding the parents' educational level, the highest percentage of children with carential rickets was registered in those with low and intermediate levels, while 54% presented a precarious socio-economic level. The prevention of carential rickets was incorrectly/incompletely performed in 38% and not performed in 19% of the cases. The average hospitalization period was 5-10 days.
Conclusions. The lack of more calcium and vitamin D in future mothers in the last trimester of pregnancy, combined with an early childbearing, precarious education, low socio-economic level, multiple births, prematurity/dysmaturity, quantitative and qualitative feeding deficits, incorrect feeding, represent risk factors within carential rickets.
Keywords: carential rickets, feeding, child

106. CRIZE CONVULSIVE IN ACIDEMIA METILMALONICA-PREZENTARE DE CAZ

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Obiectivul studiului: Se prezinta cazul unei paciente cu convulsii metabolice avand debut la varsta de 10 luni si care in urma investigatiilor s-a dovedit a fi in cadrul acidemiei metilmalonice. Acidemia metilmalonica este o boala cu transmitere autosomal recesiva care se datoreaza deficitului de metilmalonil-CoA-mutaza dependenta de vitamina B12, necesara pentru transformarea acidului malonic in acid succinic.
Materiale si metoda: Pacienta in varsta de 1 an si 6 luni a prezentat numeroase internari in spitalul nostru pentru infectii repetate, intarziere in dezvoltarea psihomotorie, mioclonii generalizate, convulsii tonice generalizate si hiperamoniemie. S-a suspicionat o boala de metabolism, ca urmare au fost dozati aminoacizii din plasma si din urina, valoare acidului metilmalonic in plasma si urina si nivelul seric al vitaminei B12.
Rezultate: Desi valorile nivelului plasmatic al vitaminei B12, aminoacizilor plasmatici serici si urinari sunt normale, nivelul crescut al acidului metil malonic in ser si urina ne orienteaza catre o forma de boala vitamina B12 non-responsiva. Desi sub tratament dietetic, cu L-carnitina, vitamina B12, acid folic, metronidazol, anticonvulsivant pacienta continua sa prezinte convulsii in cadrul crizelor metabolice.
Concluzie: In ciuda faptului ca se cunosc din ce in ce mai multe lucruri despre aceasta boala si ca diagnosticul ei precoce este posibil in tarile unde se face screeningul neonatal al acesteia, morbiditatea si mortalitatea continua sa fie ridicate. Complicatiile majore care apar sunt: afectarea pancreatica, renala, cardiaca, falimentul cresterii, atrofia nervului optic, intarzierea in dezvoltarea psihomotorie.
Cuvinte cheie: acidemie, convulsii, aminoacizi, vitamina B12.

SEIZURES IN METYLMALONIC ACIDURIA- CASE REPORT

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Purpose and objectives study: We present a girl pacient with metabolic seizures what started at the age of 10 months and after the investigations the diagnostic was Metylmalonic aciduria. Metylmalonic aciduria is a deases with autosomal recessive

which is due by the metylmalonyl CoA mutase deficit dependent on hydroxycobalamin, that is necessary for the transformation of malonic acid in succinic acid.
Materials and methods: 1 year and 6 months old female pacient with very many hospitalizations for respiratory diseases, she has delay in psychomotor development, fasciculations generalized, metabolic seizures and hyperammonaemia. We suspected a metabolic deases and for this was determined the amino acids in blood and urine, the valor of metylmalonic acid in blood and urine and the serum level of hydroxycobalamin.
Results: Although the results of hydroxycobalamin in serum is normally, amino acids in blood and urine are normally, high level of metylmalonic acid in blood and urine give us the unresponsive deases of the hydroxycobalamin. The pacient under dietery treatment, L-carnitine, hydroxycobalamin, folic acid, metronidazole and anticonvulsants therapy have many metabolic seizures with many convulsions.
Concluzions: Despite we known many thinks about this deases and its early diagnosis is posibil in countries where screening is done, mortality and morbidity remains high. Major complications are occurring: damage in pancreas, kidney, heart, growth failure, optic nerve atrophy, delay in psychomotor development
Keys: aciduria, amino acids, seizures, hydroxycobalamin.

ENDOCRINOLOGIE ŞI DIABET PEDIATRIC / PEDIATRIC ENDOCRINOLOGY AND DIABETES

107. ABORDAREA NOU NASCUTULUI CU VARSATURI - ASPECTE PRACTICE

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Introducere : Varsatura este unul din cele mai frecvente simptome din patologia pediatrica, cauza unor tulburari motorii, metabolice, nervoase sau obstructive la diferite niveluri ale tractului digestiv sau al altor organe, administrarii unor medicamente, a unor substante toxice sau a unor stari fiziologice.
Prezentare de caz: Prezentam cazul unui nou-nascut în varsta de 26 zile, internat in Clinica Pediatrie I Targu-Mures pentru varsaturi postalimentare, unele in jet, apetit capricios, scadere in greutate, somnolenta. Sub tratamentul complex instituit starea sugarului a fost lent favorabila, cu episoade de usoara ameliorare, urmate de recaderi, iar examenele de laborator efectuate (hemocultura, urocultura, coprocultura, latex-Rotavirus) nu au putut duce initial la stabilirea unui diagnostic cert. S-au suspicionat reflux gastroesofagian, sindrom de colestaza, stenoza hipertrofica de pilor, intoleranta la lactoză. Serologia TORCH a fost negativa.
Deoarece se mentine hiponatremia, hiperkaliemia sub tratament cu electroliti intravenos, s-a ridicat suspiciunea unui sindrom adrenogenital congenital cu deficit de 21 hidroxilaza cu pierdere de sare, motiv pentru care s-au recoltat 17 OH progesteron, cortizolemia, care au pus in evidenta valori patologice. Evolutia nefavorabila a necesitat tratament medical prompt.
Concluzii : Varsatura este un semn de falsa banalitate, atat prin faptul ca poate constitui semnalul unor afectiuni severe cat si prin consecintele sale imediate (deshidratare, acidoza, colaps) sau pe termen lung (distrofie, pericol de aspiratie, si chiar deces).
Cuvinte cheie : varsatura, nou nascut, hiponatremie

A PRACTICAL APPROACH OF THE NEWBORN WITH VOMITING

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Introduction: Vomiting is one of the most common symptoms in pediatric pathology, caused by a motor, metabolic or nervous disorder, an obstruction at different levels of the digestive sistem or of other organs, consequence of drugs or toxic substances administration, or it can be a physiological fact.
Case presentation: We present the case of a 26 days old newborn, admitted in our Clinic, Pediatrics 1 Tirgu Mures, for postalimentar vomiting, which some were projectile, poor feeding, weight loss and drowsiness. With the complex treatment initiated the newborn status was slowly favorable, with episodes of slight improvement, followed by relapse, while the laboratory investigations performed (blood, urine and stool culture, latex-Rotavirus) couldn't lead initially to establish a

definite diagnosis. Was suspected a gastroesophageal reflux, cholestasis syndrome, hypertrophic pyloric stenosis, lactose intolerance.TORCH serology was negative.
Because hyponatraemia and hyperkalaemia were persistent under intravenous treatment with electrolytes, we suspected a congenital adrenogenital syndrome with 21-hydroxylase deficiency and salt loss, reason why we determined 17-OH progesterone, cortisolemia, which showed pathological values. This unfavorable status requested prompt medical treatment.
Conclusions: Vomiting is a sign of false triviality, so that it can be the signal of severe disease and by its immediate consequences (dehydration, acidosis, collapse) or long term (dystrophy, risk of aspiration, and even death).
Key words: vomiting, newborn, hyponatremia

108. ABORDAREA HIPOGLICEMIEI SEVERE PERSISTENTE LA COPIL

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Hipoglicemia la copil poate fi definita ca un grup de tulburari ce reflecta esecul mentinerii homeostaziei normale a glucozei, determinat fie de deficienta substratului (depozite insuficiente de glicogen), fie de deficienta enzimelor si hormonilor, implicati in metabolismul hidrocarbonatelor. In copilarie, hipoglicemia persistenta poate avea un impact major asupra dezvoltari structurale si functionale a creierului, de aceea se impune stabilirea unui diagnostic si tratament cat mai precoce. Ne propunem sa prezentam o serie de cazuri clinice de hipoglicemie severa la copil , care au ridicat probleme de diagnostic. Plecand de la aceste cazuri vom prezenta o schema de abordare diagnostica a hipoglicemiilor severe persistente la copil. In concluzie, desi majoritatea cazurilor de hipoglicemie la copil apar prin post prelungit exista si alte cazuri de hipoglicemie severa persistenta, diagnosticul etiologic al acesteia devenind astfel dificil si impunand o abordare complexa. Cuvinte cheie : hipoglicemie severa,persistenta; copil.

PERSISTENT SEVERE HYPOGLYCAEMIA APPROACH IN CHILDREN

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Hypoglycemia in children can be defined as a group of disorders that reflects failure to maintain normal glucose homeostasis, due to either substrate deficiency (insufficient storage of glycogen) or enzyme deficiency and hormones involved in metabolism of carbohydrates. In childhood , persistent hypoglycemia can have a major impact on the structural and functional development of the brain , so it requires a diagnosis and a treatment as quickly as possible. We intend to present a series of clinical cases of severe hypoglycemia in children , raising diagnostic problems . Considering these cases we present a schematic approach to diagnose severe persistent hypoglycemia in children. In conclusion, although most cases of hypoglycemia in children occur through prolonged starvation, there are other cases of persistent severe hypoglycemia making its etiologic diagnosis difficult and imposing a comprehensive approach.

109. EVALUAREA RISCULUI CARDIOVASCULAR LA COPIII OBEZI PRIN METODE ANTROPOMETRICE, BIOLOGICE ŞI IMAGISTICE

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Obezitatea în randul copiilor este în continua crestere, fiind diagnosticata la varste tot mai mici. Scopul studiului: Evaluarea parametrilor clinico-biologici si a modificarilor care apar la copiii cu obezitate; identificarea sindromului metabolic (SM) in cadrul loturilor studiate; identificarea, evaluarea, analiza si corelatia factorilor adipogeni cu indicele intima medie carotidian (IMC). Material si metoda: Un studiu crossectional a fost efectuat pe o perioada de 1 an (aprilie 2014-aprilie 2015). Acesta a cuprins 68 pacienti cu obezitate cu varsta medie 11,83 ani distribuiti astfel: 17 (25%) au avut varsta cuprinsa intre 5 si 9, 35 (51%) intre 10 si 14, iar 16 (24%) intre 15 și 18 ani. Tensiunea arteriala, lipidograma, glicemia, leptina, adiponectina si CRP inalt specific au fost determinate. S-a efectuat testul de toleranta la glucoza orala. Rezistenta la insulina (IR) a fost evaluata prin HOMA. IMC a fost masurat la toti pacientii. Rezultate: SM a fost prezent la 18 (26,14%) din pacienti, cu o prevalenta crescuta in randul grupului 15-18 ani (11.76% vs. 22.85% vs.50%). S-a putut observa o corelatie puternica intre IMC si ceilalti factori metabolici (r=0.83). Adiponectina, leptina, CRP inalt specific, IMC au valori mult mai mari la grupul de varsta 15-18 ani (p=00.4,p=0.035, p=0.03). Concluzie: Riscul metabolic creste o data cu varsta. Exista o corelatie puternica intre IMC si adiponectina, leptina, CRP-inalt specific. IMC este un cunoscut marker subclinic pentru ateroscleroza, este o metoda ieftina si noninvaziva. Studii populationale extinse sunt necesare in vederea stabilirii unor valori prag în ceea ce priveste IMC.
Cuvinte cheie: risc cardiovascular, obezitate, copil

ANTHROPOMETRIC, BIOLOGICAL AND IMAGISTICAL METHODS FOR ASSESSING THE CARDIOVASCULAR RISK IN OBESE CHILDREN

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Pediatric obesity has increased worldwide over the last decades, being diagnosed at ever-younger ages. Aim: Evaluation of clinical and biological parameters and changes that occur in children with obesity; metabolic syndrome (MetS) identification in the studied groups; identification, evaluation, analysis and correlation of the adipogenic factors with the carotid intima media thickness (CIMT). Material and methods: A cross-sectional study was conducted over a period of 1 year (April 2014-april 2015). 68 obese patients with mean age 11.83 years were included, distributed as follows: 17 (25%) were aged between 5 and 9, 35 (51%) between 10-14 and 16 (24%) between 15 and 18. Blood pressure, lipids, glucose, leptin, adiponectin and high sensitive CRP were determined. Oral glucose tolerance test was performed in all children. Insulin resistance (IR) was assessed by HOMA. CIMT was measured in all patients. Results: MetS was present in 18 patients (26.14%), with a higher prevalence among the 15-18 age group (11.76% vs. 22.85% vs.50%). A strong correlation between CIMT and other metabolic factors has been observed (r = 0.83). Lower levels of adiponectin, higher levels of leptin, high sensitive CRP and CIMT values have been observed on the 15-18 age group (p = 00.4, p = 0.035, p = 0.03). Conclusions: Metabolic risk increases with age. There is a strong correlation between CIMT and adiponectin, leptin, high sensitive- CRP. CIMT is a known marker for subclinical atherosclerosis, it is a cheap and noninvasive method. Extensive population studies are required to establish threshold values for CIMT in children.

Key words: cardiovascular risk, obesity, child

110. UN SINDROM DE COLESTAZA LA UN PACIENT CU PARALIZIE CEREBRALA SI EPILEPSIE IN TRATAMENT

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Prezentare de caz: Gabriela, 8 ani, s-a internat in Clinica I Pediatrie, Timisoara pentru elucidarea unui sindrom de colestaza, evidentiat la analizele de rutina de medicul de familie. Este cunoscuta cu Paralizie cerebrala forma hipotona, Epilepsie cu crize polimorfe partial controlate medicamentos cu Depakine, Fenitoina, Levomepromazin si 6 fracturi ale membrelor inferioare produse in ultimii 3 ani in timpul crizelor.

In momentul internarii, tegumentele erau palide, cu tesut subcutanat absent si atrofie musculara. Prezenta atela gipsata la nivelul membrului inferior stang si pseudoartroza in 1/3 distala a gambei drepte. Era echilibrata cardio-pulmonar, cu abdomen moale, alimentata pe sonda nazo-gastrica, tranzit lent si diureza prezenta.

Investigatii biologice au evidentiat TGP=11U/l, TGO=20U/l, GGT=46U/l, FA=2980U/l, prezenta de amidon si fibre in scaun, in rest valori normale. Evaluarea metabolismului fosfo-calcic a surprins Ca^{2+}=0.81mmol/l, Catotal=1.67mmol/l, 25-OHvitaminaD=16.0µg/l, intactPTH=613.9pg/ml, βcrosslaps>6ng/ml, osteocalcina=78.86ng/ml si fosfataza alcalina osoasa=1174.0µg/L.

Radiografiile de umar si gamba bilateral au evidentiat leziuni de osteoliza si vechi fracturi de clavicula stanga, 1/3 superioara diafiza humerus si femur stang si 1/3 inferioara diafiza tibie dreapta, vindecate vicios, pe fond de osteogeneza imperfecta.

S-a stabilit diagnosticul de rahitism sechelar prin deficit de vitamina D, prin tratament cronic cu anticonvulsivante si maldigestie/malabsorbtie, hiperparatiroidism secundar si osteogeneza imperfecta.

Evolutia a fost favorabila sub tratament cu calciu gluconic (100mg/kg/zi iv), vitaminaD (10 picaturi/zi), cu reducerea valorilor FA=1500U/l la externare. S-a luat in calcul initierea tratamentului cu bifosfonati.

Particularitatea cazului: Asocierea rahitismului sechelar si a osteogenezei imperfecta-tip IV in producerea fracturilor la un pacient cu paralizie cerebrala si epilepsie.

Cuvinte cheie: rahitism carential, osteogeneza imperfecta, epilepsie

CHOLESTASIS SYNDROME IN A PATIENT WITH CEREBRAL PALSY AND EPILEPSY WITH TREATMENT

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Case presentation: Gabriela, 8 years old, was admitted in the 1st Pediatric Clinic, Timisoara for the evaluation of cholestasis syndrome. She was known with hypotonic cerebral palsy, epilepsy with polymorphic seizures partial controlled by Depakine and Phenytoin and 6 leg fractures occurred in the last 3 years during crises.

At admission, she had pale skin, absent subcutaneous tissue and muscle atrophy. She had splint cast in the left lower limb and pseudoarthrosis in the 1/3 distal of the right leg. Cardiopulmonary was normal with painless abdomen and feed by nasogastric tube. Her intestinal transit was slow and the diuresis was presented.
The biological investigations revealed TGP=11U/l, TGO=20U/l, GGT=46U/l, FA=2980U/l, the presence of starch and fiber in the stool, while the other tests were normal. The phosphor-calcium metabolism surprised the presence of $Ca2^{+}$=0.81mmol/l, totalCa=1.67mmol/l, 25-OH vitaminD=16.01µg/l, intactPTH=613.9pg/ml, βcrosslaps> 6ng/ml, osteocalcin=78.86ng/ml and bone alkaline phosphatase =1174µg /l.
The radiography of shoulders and legs showed osteolysis injuries and old left clavicle, upper third diaphysis of left humerus and femur and lower third diaphysis of right tibia fractures, with vicious callus secondary to Osteogenesis Imperfecta.
The diagnoses were rickets due to vitaminD deficiency, chronic anticonvulsant therapy and malabsorption syndrome and Osteogenesis Imperfecta.
The evolution was favorable under treated with calcium gluconate (100mg/kg/day IV), Vigantol (10drops /day) with the reduction of FA=1500U / l.The initiation of bisphosphonate therapy was taken into account.
Case particularity: The association of rickets and Osteogenesis Imperfecta responsible for the production of fractures in a patient with cerebral palsy and epilepsy.
Key words: rickets, osteogenesis imperfecta, epilepsy

111. BOALA CELIACA SI DIABETUL DE TIP 1 LA COPIL – STUDIU CLINC

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Boala celiacă definita ca intoleranţa permanentă la gliadine şi proteinele înrudite din unele cereale (grâu, orz, ovăz, secară) determina un răspuns imun anormal la nivelul mucoasei intestinale la indivizii susceptibili genetic. Un fond genetic comun asociat cu hiperexpresia antigenelor HLA DQ2/DQ8 și un mecanism patogenetic similar în declanșarea procesului autoimun conduce la o prevalența bolii celiace la pacienții cu diabet zaharat tip 1 (Dz1) de aproximativ 10 ori mai mare decât în populația generală. Studiu descriptiv retrospectiv realizat pe 3 ani, lot de 180 de copii cu varsta intre 3-18 ani, cu Dz1, aflati in evidenta compartimentului de “Diabet zaharat si Boli de nutritie” al Sp. “ M.S. Curie” – Bucuresti. S-a urmarit asocierea Dz 1 cu boala celiaca: dozare Ac anti transglutaminaza (Ac anti TG), Ac antiendomisium (EMA), efectuare biopsie jejunala, evaluare echilibrul glicemic (dozare HbA1c). Rezultate: din 180 de pacienti cu Dz 1 la care s-au dozat Ac anti TG, EMA, 28 au avut valori mult peste limita normala a Ac anti TG; 10 dintre pacienti- biopsia jejunala a confirmat celiachia - stad. Marsh III; restul pacientilor au prezentat valori ale Ac anti TG > 20U/ml si < 40-60U/ml. In concluzie diagnostigarea bolii celiace si aplicarea dietei de excludere la copii cu Dz 1 influenteaza in mod pozitiv dezvoltarea acestora si imbunatateste controlul glicemic. Cuvinte cheie: celiachie, diabet, autoimunitate

CELIAC DISEASE AND TYPE 1 DIABETES IN CHILDREN – CLINICAL STUDY

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Celiac disease defined as permanent intolerance to gliadin and related proteins in some cereals (wheat, barley, oats, rye) cause an abnormal immune response in the gut in genetically susceptible individuals. A common genetic background associated with HLA DQ2/ DQ8 hyperexpression and a similar pathogenic mechanism in triggering the autoimmune process leads to a prevalence of celiac disease in patients with type 1 diabetes of about 10 times higher than in the general population. Retrospective descriptive study conducted on 3years period, on 180 children aged 3-18 years diagnosed with type 1 diabetes, admitted in the " Diabetes and Nutrition diseases " compartment of "M.S. Curie " Hospital from Bucharest . It was followed the associationof between type 1 diabetes and celiac disease: determination of Ac anti transglutaminase (anti- TG) , Ac antiendomisium (EMA), glycemic control (HbA1c dosage). Results : from 180 patients with type 1 diabetes that were dosed TG -Ab, EMA, 28 had values well above normal TG - Ab; jejunal biopsy of 10 of patients- confirmed celiac disease - stage Marsh III ; the remaining patients had values of TG -Ab > 20U / mL and < 40-60U / ml. In conclusion diagnosis for celiac disease and the application of exclusion diet in children with type 1 diabetes positively influences their development and improves glycemic control. Keywords : celiac disease , diabetes, autoimmunity

112. PREVALENTA BOLILOR ALERGICE LA COPIIICU DIABET ZAHARAT TIP 1

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Diabetul Zaharat tip 1 este considerat o boala autoimuna mediata de limfocitele Th1, in timp ce bolile alergice se caracterizeaza prin raspuns imun mediat de Th2.Exista numeroase controverse referitoare la asocierea sau excluderea reciproca a acestor maladii, insa paradigma clasica a excluderii reciproce a bolillor mediate de Th1, respectiv de Th2, a fost revizuita luand in considerare atat rolul celulelor reglatoare Threg, dar si al factorilor de mediu implicati.

Obiective:Scopul studiul este investigarea frecventei bolilor alergice (rinita, astm bronsic, dermatita) la copii diagnosticati cu cu DZ1.

Material si Metode: Studiul retrospectiv a inclus 101 pacienti cu DZ1 (46 fete si 55 baieti) aflati in observatia Clinicii Pediatrie I Cluj-Napoca in perioada 2010-2014, cu varste cuprinse intre 4 si 19 ani. S-a determinat prevalenta bolilor alergice, nivelul IgE, calitatea controlului glicemic, evaluat prin HbA1c, la cei cu DZ1 si alergii (lot I: 21 pacienti) si lotul fara alergii (lot II: 80 pacienti).

Rezultate: Din cei 101 pacienti cu DZ1, 21 pacienti (20,79%) au asociat DZ1 cu boli alergice. Nivelul IgE a fost superior intervalului de referinta corespunzator varstei la 38,09% din lotul I.

47,7% dintre pacientii lotului I au prezentat HbA1c peste 7,5%, comparativ cu 30,2% dintre pacientii lotului II.

Concluzii: Asocierea DZ1 cu bolile alergice este relativ frecventa (unu din cinci), iar calitatea controlului glicemic a fost moderat influentata de asocierea cu bolile alergice.

Cuvinte cheie: diabet zaharat tip1, rinita alergica, astm, imunoglobulina E, copii

THE PREVALENCE OF ALLERGIC DISEASES IN CHILDREN WITH TYPE 1 DIABETES MELITUS

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Diabetes mellitus type 1 (DM1) is considered an autoimmune disease mediated by Th1 lymphocytes, while allergic diseases are characterized by Th2-mediated immune response. There are many controversies about the association or mutual exclusion of these diseases, but classical paradigm based on the assumption that diseases mediated by Th1 and Th2 should be mutually exclusive, has been revised considering both the role of regulatory cells Threg, and the environmental factors involved.

Objectives:The aim of this study was to investigate the frequency of allergic diseases (rhinitis, asthma, dermatitis) in children diagnosed with DM1.

Material and Methods: The retrospective study included a total of 101 patients with DM1 (46 girls and 55 boys aged 4 - 19 years) followed at the Pediatric Clinic I Cluj-Napoca in 2010-2014. We determined the prevalence of the allergic diseases, IgE level, quality of glycemic control as measured by HbA1c in patients with DM1 and allergies (group I) and without allergy group (group II).

Results: Of the 101 patients with DM1, 21 patients (20.79%) DM1 associated with allergic diseases. IgE levels were higher than age appropriate reference range for 38.09% of patients in group I.

47.7% of patients in group I had HbA1c above 7.5%, compared with 30.2% of patients in group II.

Conclusions:In the studied group, the associaton of DM1 with allergic diseases is relatively frequent (one of five) and the quality of glycemic control was moderately influenced by this association.

Keywords: type 1 diabetes mellitus, allergic rhinitis, asthma, Imunglobulin E, children

113. EVALUAREA ETIOLOGIEI DEFICITULUI STATURAL LA COPII

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Introducere

Statura mica reprezinta o provocare diagnostica pentru clinicieni. De multe ori, diagnosticul este intarziat de lipsa sistematica de evaluarea a cresterii staturale la copii.

Obiective

Ne-am propus sa evaluam retrospectiv etiologia deficitului statural la copiii care s-au pezentat pentru evaluari diagnostice ale staturii mici.

Material si metoda

In Clinica de Pediatrie Constanta (ianuarie 2014-mai 2015), a fost evaluat deficitul statural a 55 de copii. Evaluarea clinica: masurarea inaltimii, greutatii, pozitionarea pe graficele de crestere, scorul Prader, viteza de crestere, radiografie de pumn, radiografie sa turcica, CT sau RMN hipotalamo-hipofizar. A fost evaluata functia tiroidiana la toti pacientii. In functie de aspectul clinic si anamneza s-au efectuat: cariotip, testul sudorii, test celikey, alte investigatii de laborator.

Deficitul de hormoni de crestere a fost evaluat cu ajutorul testelor de stimulare cu insulina si arginina.

Rezultate
15% au fost diagnosticati cu intarziere constitutionala a cresterii, 19 % malnutritie, 35% boli cronice, 10% sindroame genetice (Silver-Russel, Turner), 21% hipotiroidism, 6% prematuri,9 % SGA.
Varsta medie de diagnostic a deficitului statural este de 6.86 ani pentru nanismul hipofizar, 6.9 ani pentru malnutritie si 8.56 ani pentru sindromul Turner.
Intarzierea varstei osoase, comparativ cu varsta cronologica, a prezentat o medie de 2,3 ani.
Testele de stimulare a secretiei hormonului de crestere s-au efectuat la 35 pacienti. La 42% valorile GH post stimulare au fost sub 5 ng/ml (normal >5).
Concluzii
Evaluarea cresterii si dezvoltarii trebuie efectuata anual. Diagnosticarea tardiva a deficitului de crestere limiteaza posibilitatile terapeutice de corectare.
Cuvinte cheie: deficit statura sindrom deviere varsta

EVALUATION OF SHORT STATURE ETIOLOGY IN CHILDREN

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Introduction
Short stature is a challenge for clinicians to diagnose. Often, the diagnosis is delayed by the lack of systematic evaluation of stature in children`s growth.
Objectives
We aimed to evaluate retrospectively the etiology of stature deficiency in children who were submitted for diagnostic evaluation of short stature.
Methods
In Pediatric Clinic of Constanta (January 2014- May 2015) we evaluated stature deficiency in 55 children. Clinical evaluation: measurement of height, weight, growth charts positioning, Prader score, growth speed, hand x-ray, pituitary gland x-ray, hypothalamic-pituitary CT or MRI.Thyroid function was evaluated in all patients. Depending on the clinical appearance and history the following were performed: karyotype, sweat test, Celiac disease test, and other laboratory investigations. Deficiency of growth hormone was evaluated using stimulation tests with insulin and arginine.
Results
15% were diagnosed with constitutional delay of growth, 19% with malnutrition, 35% with chronic diseases, 10% with genetic syndromes (Silver-Russell, Turner), 21% with hypothyroidism, and 6% with prematurity, 9% with SGA. The average age of diagnosing stature deficiency is 6.86 years for pituitary dwarfism, 6.9 years for malnutrition and 8.56 years for Turner syndrome. Delayed bone age compared with chronological age, presented an average of 2.3 years. Tests to stimulate growth hormone secretion were performed in 35 patients. At 42% GH values after stimulation were below the 5 ng / ml.
Conclusion
An evaluation of growth and development should be performed annually. Late diagnosis of growth deficiency limits therapeutic possibilities for correction.
Keywords: deficit stature syndrome deviation age

PNEUMOLOGIE / PNEUMOLOGY

114. ASPERGILOZA BRONHOPULMONARA ALERGICA - AFECTIUNE RAR DESCRISA LA VARSTA PEDIATRICA

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Aspergiloza bronhopulmonara alergica (ABPA) este o boala pulmonara mai frecvent intalnita in randul adultilor si rar intalnita la varsta pediatrica. Este prezentat cazul unei paciente in varsta de 15 ani 9 luni cunosuta de la varsta de sugar cu episoade de tuse, respiratie dificila si wheezing interpretate ca bronsiolita

acuta, pneumonie acuta sau astm bronsic. A prezentat numeroase astfel de episoade, de intensitate medie sau severa, necesitand spitalizare repetata. In ianuarie 2014 este internată într-un alt seviciu medical cu o exacerbare severă care a impus corticoterapie orală de lungă durată cu Medrol. Investigatiile efectuate releva un titru IgE mult crescut (6882 UI). Întrucât se menține cu dispnee, cu necesar de cortizon și oxigenoterapie a fost internată în Clinica Pediatrie III. In aceste conditii s-au efectuat investigatii de laborator ce confirma IgE totale 2500 UI, un titru moderat al IgE specifice pentru Aspergillus fumigatus, iar probele functionale respiratorii confirma obstructie bronsica cu disfunctie ventilatorie severa de tip obstructiv, responsiva la administrarea de ventolin. Examenul CT toracic releva modificari bronho-pulmonare sugestive pentru aspergilloza bronhopulmonara alergica. Asociat pacienta prezinta modificari ECG de tipul hipertrofiei atriale drepte, suprasolicitarea ventriculului drept cu deviere axiala dreapta importanta. Ultrasonografie cardiaca pune in evidenta dilatarea cavitatilor drepte, sugestive pentru cord pulmonar cronic. S-a initiat terapie specifica cu lantifuncide și s-a continuat corticoterapia sistemica si inhalatorie asociata cu beta2 agonisti de lungă durata, cu ameliorarea simptomelor și a parametrilor funcționali respiratori precum si a celor imunologici la 3 luni de la diagnostic.

ALLERGIC BRONCHOPULMONARY ASPERGILLOSIS – A VERY RARE CONDITION DESCRIBED AT PEDIATRIC AGE

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Allergic bronchopulmonary aspergillosis (ABPA) is a lung disease most common among adults and rarely seen in the pediatric age. We present the case of a patient about 15 years 9 months known from infant age with numerous episodes of cough, wheezing and shortness of breath interpreatate like acute bronchiolitis, acut pneumonia or acute asthma. She presented many such episodes moderate or severe annually, requiring repeated hospitalization. In January 2014 she is hospitalized in another hospital with a severe exacerbation that imposed on long-term oral corticosteroids with Medrol. Investigations made have revealed increased IgE titers (6882 IU). As she is maintained with dyspnea, cyanosis, with required systemic cortisone and oxygen-terapy she was hospitalized in our clinic - Pediatrie III. In these circumstances laboratory investigations have confirmed IgE Total 2500 IU, moderate IgE specific titers for Aspergillus fumigatus. Spirometry confirmed bronchial obstruction with severe obstructive ventilatory dysfunction that responded at Ventolin administration. Chest CT was performed and the broncho-pulmonary changes where included in allergic bronchopulmonary aspergillosis. Associated our patient present ECG changes like right atrial, hypertrophy, right ventricular overload with important right axial deviation. Cardiac ultrasonography reveals the right cavity dilation, suggestive for chronic pulmonary heart. It was initiated the terapy with antifungal and was continued systemic and inhaled corticosteroid therapy associated with long-term beta2 agonists, with the improvement of symptoms, functional respiratory parameters and immunological tests at 3 months after diagnosis.

115. VALIDAREA UNUI SCOR CLINIC DE TRAIAJ SI RASPUNS LA TRATAMENTUL INITIAL AL COPIILOR CU SINDROM BRONHO-OBSTRUCTIV ACUT

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Recunoasterea, triajul rapid si initierea primelor masuri terapeutice aplicate copiilor cu bronhospasm pot fi salvatoare de viata.

Pentru ameliorarea rulajului acestor pacienti autorii prezenta un scor clinic simplu folosit in CPU. Evaluarea este rapida si simpla, fiind usor de efectuat in lipsa oricaror mijloace paraclinice. Pulmonary Index Score [PIS] are patru parametri clinici, distribuiti in 4 clase de severitate. Acestia sunt frecventa respiratorie, durata raportului inspir/expir, intensitatea wheezingului si folosirea muschilor respiratori accesorii. Copii cu sindrom bronhoobstructiv acut pot fi impartiti in trei clase de severitate: usor, mediu sau sever, cu scor de la 0 la 12. Folosirea acestei abordari, alaturi de alte masuri organizatorice, au dus la cresterea semnificativa a rulajului copiilor cu afectiuni acute in CPU. Comparativ cu 2008 s-a observant in iarna 2014/15 o crestere a prezentarilor de 3 ori, iar a proportiei pacientilor cu bronhospasm de la 15,24% la 16,8% total prezentari [chi^2 = 39.6906, p<0,00001]. Din cei 1044 copii cu bronchospasm evaluati in lunile decembrie 2014 si ianuarie 2015 au fost analizati 134 pacienti cu o varsta medie de 34,6 luni, o severitate usoara/medie [media scor PIS=4,33]. 14,4% au avut indicatie de internare si 8,33% s-au internat efectiv. Din cei 121 copii (91,67%) care nu s-au internat, imensa majoritate (113 = 93,4%) au fost tratati in CPU si trimisi la domiciliu dupa stabilizare.

In concluzie folosirea PIS creste semnificativ statistic eficienta triajului copiilor cu dispnee expiratorie in UPU/CPU, in special in sezonul hibernal, cand 1/6 prezentari constituie astfel de pacienti.

CUVINTE CHEIE – pulmonary index score, triaj, bronhospasm, copil

VALIDATION OF A CLINICAL SCORING SYSTEM USED FOR TRIAGE AND EMERGENCY TREATMENT RESPONSE IN CHILDREN WITH ACUTE BROCHOSPASM

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Rapid assessment of airway and timely action can prevent deterioration of respiratory failure to respiratory arrest. In a busy emergency department (ED) rapid triage can be performed with a relative simple clinical-scoring system. Pulmonary Index Score can be performed without any laboratory testing, so it is a rapid tool for ED.

Four parameters [respiratory rate, wheezing, inspiration/exhalation time ratio and accessory muscle use] are quantified in four severity classes rated from 0 to 3. Severity of bronchial obstruction can be evaluated via PIS as none, mild, moderate or severe, with a score from 0 to 12. During winter 1 in 6 children presented to our ED had bronchospasm. Authors present the impact of a structured approach based on PIS. This strategy is efficient [chi^2 = 39.6906, $p<0.00001$] and decreases the admission rate of these children. A subgroup of 134 patients is analyzed. Average age is 34.6 months and average PIS is 4.33. Most of the children were treated as outpatients in ED – 93.4%.

Conclusion – Pulmonary Index Score is a valuable and efficient tool in ED, mainly during cold season.

KEY WORDS – pulmonary index score, triage, bronchospasm, child

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116. ASTMUL IN FIBROZA CHISTICA

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Fibroza chistica (FC) este cea mai comuna afectiune autozomal recesiva la populatia caucaziana. Boala pulmonara se caracterizeaza prin afectarea clearance-ului mucociliar cu obstructia cailor aeriene, infectie si inflamatie cronica. Wheezingul esta un simptom comun în FC si în timp ce la cei mai multi pacienti este determinat de boala pulmonara subiacenta, în unele cazuri wheezingul e determinat de prezenta concomitenta a astmului. Este dificil sa determinam care pacienti au combinatie cu astmul si care au simptome asemanatoare astmului, determinate de inflamatia plamânului din FC. Nu este un consens în cum definim astmul în fibroza chistica, dar diagnosticul se bazeaza în principal pe istoricul personal si familial de atopie, al pacientului. Testarea functiei pulmonare (spirometrie, reversibilitate la bronhodilatator, hiperreactivitatea bronsica), excluderea altor diagnostice (refluxul gastroesofagian, traheobronhomalacia, ABPA, bronsiolita obliteranta) si raspunsul la medicatia antiastmatica, pot ajuta la diagnostic. Identificarea pacientilor cu astm concomitent semnificativ este totusi importanta, deoarece pot beneficia de medicatia specifica astmului, în special corticosteroizi inhalatori. Indiferent daca wheezingul este datorat unui astm concomitent sau nu, tratamentul urmeaza progresia standard, in trepte, folosita în astm.

Cuvinte cheie: fibroza chistica, wheezing, astm, atopie

ASTHMA IN CYSTIC FIBROSIS

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Cystic fibrosis (CF) is the most common autosomal recessive disease in Caucasians. Lung disease is characterized by impaired mucocilliary clearance with airway obstruction and chronic pulmonary infection and inflammation. Wheeze is a common symptom in CF and, whilst in many patients this is due to their underlying lung disease, in some cases the wheeze is due to the presence of concomitant asthma. It is difficult to determine which patients have a combination of asthma and which have asthma like symptoms caused by inflammation of the CF lung. There is no consensus on how to define CF asthma, but the diagnosis is predominantly based on the patient’s strong family and personal history of atopy. Pulmonary function tests (spirometry, bronchodilator responsiveness, BHR), other diagnose excluded (gastroesophageal reflux, malacic airways, ABPA, obliterative bronchiolitis) and the response to antiasthma medication, may also help the diagnosis. Identifying patients with significant co-existing asthma is, however, important as they may benefit from specific asthma medications, in particular inhaled steroids. (11) Whether wheezing is due to concomitant asthma or not, the treatment follows the standard step-wise progression used in asthma.

Key words: cystic fibrosis, wheeze, asthma, atopy

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117. COMPLCATII PULMONARE NEINFECTIOASE IN BOALA RESPIRATORIE DIN FIBROZA CHISTICA

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Fibroza chistica este o boala multisistemica complexa, cauzata de defectul intr-o singura gena. Manifestarile pulmonare sunt responsabile pentru cea mai mare morbiditate si mortalitate. Odata ce boala pulmonara progreseaza, exista un risc crescut de complicatii respiratorii. Atelectazia lobara poate fi asimptomatica si remarcata doar in momentul unei radiografii toracice de rutina. Colapsul pulmonar sau lobar este o complicatie relative rara a fibrozei chistice, iar antibioterapia agresiva, rh-DN aza,

solutia salina hipertona si cresterea fizioterapiei toracice, poate fi eficienta. Daca acestea esueaza, bronhoscopia trebuie efectuata. Hemoptizia si pneumotoraxul sunt complicatii care apar in boala pulmonara avansata. Hemoptizia minora este comuna in fibroza chistica. Sangerarea endobronsica este consecinta eroziunii peretelui cailor aeriene secundara inflamatiei si infectiei. Hemoptizia majora este de obicei asociata cu boala severa si poate fi controlata prin embolizarea arterei bronsice. Pneumotoraxul poate fi o complicatie grava a pacientilor mai in varsta. Un pneumotorax mic este abordat conservator, dar un pneumotorax mare sau sub tensiune, necesita rapid tratament de drenaj. In boala pulmonara usoara, hipoxia este principala anomalie a schimbului de gaze (insuficienta respiratorie tip1). Odata cu progresia bolii, tipul 2 de insuficienta respiratorie cu retentie de CO2 survine eventual. Insuficienta cardiaca dreapta cronica (cordul pulmonar) este o complicatie observata la pacientii cu CF si boala pulmonara avansata. Cordul pulmonar este recunoscut la pacientii cu hipoxie si hipertensiune pulmonara.

cuvinte cheie: fibroza chistica, atelectazie, hemoptizie, pneumotorax, insuficienta respiratorie

NON-INFECTIOUS COMPLICATIONS IN RESPIRATORY DISEASE FROM CYSTIC FIBROSIS

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Cystic fibrosis(CF) is a complex multisystem disease, caused by defects in a single gene. Pulmonary manifestations are responsible for most of the morbidity and mortality. With pulmonary disease progression, there is an increased likelihood of respiratory complications. Lobar atelectasis may be asymptomatic and noted only at the time of a routine chest radiograph. Lung or lobar collapse is a relatively rare complication of CF and aggresive antibiotic therapy, rhDNase, hypertonic saline and increased chest physiotherapy may be effective. If this fails bronchoscopy should be performed. Haemoptysis and pneumothorax are complications in advanced lung disease. Minor haemoptysis is common in CF and more so in older patients. Endobronchial bleeding is the consequence of airway wall erosion secondary to inflammation and infection. Major haemoptisys is usually associated with severe disease and can be controlled by bronchial artery embolisation. Pneumothorax may be a life-threatening complication in older patients. A small pnumothorax is managed conservatively but a large pneumothorax or under tension requires rapid treatment with drainage. In mild CF lung disease, hypoxia is the main gas exchange abnormality (type 1 respiratory failure). As disease progresses, type 2 respiratory failure with CO2 retention eventually supervenes. Patients progress to chronic respiratory failure from slow deterioration of lung function. Chronic right sided heart failure (or pulmonale) is a complication seen in CF patients with advanced pulmonary disease. Cor pulmonale is well recognized in patients with hypoxia and pulmonary hypertension.

key words: cystic fibrosis, atelectasis, haemoptysis, pneumothorax, respiratory failure, cor pulmonale

118. MANAGEMENTUL INFECTIEI PULMONARE LA PACIENȚII CU MUCOVISCIDOZA (FIBROZA CHISTICA) : UP-TO-DATE

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Introducere: Fibroza chistica (mucoviscidoza) este cea mai frecventa boala monogenica, caracterizata printr-un polimorfism clinic marcat,al carei evolutie este dictata de afectiunea pulmonara,care constituie elementul principal in prognosticul bolnavului cu fibroza chistica (FC).

Infectiile bacteriene sunt responsabile pentru majoritatea exacerbarilor la pacientii cu FC , etiologia fiind frecvent polimicrobiana si cu suse rezistente. Germenii „traditional” asociati FC sunt: *Pseudomonas aeruginosa, Staphylococcus aureus, Haemophilus influenzae,* in timp ce *Burkholderia cepacia complex, Stenotrophomonas maltophilia, Achromobacter xylosoxidans* sunt mai nou incriminati. Statusul pulmonar este influentat de numarul si gravitatea exacerbarilor, iar evoluţia bolii este marcata de momentele infecţioase , care duc la pierderea progresiva a functiei pulmonare, spre insuficienta respiratorie cronica. Acutizarile respiratorii duc la pierderea functionala si organica a parenchimului pulmonar, primoinfectia cu Pseudomonas fiind frecvent urmata de colonizare. Prezervarea functiei pulmonare este principalul scop al terapiei in FC, deoarece, o data pierduta , ea nu mai poate fi recuperata. Antibioterapia trebuie instituita precoce, in doza si timp adecvate, conform consensurilor actuale si in conjunctie cu alte metode terapeutice specifice ca fizioterapia .

In concluzie: Interventia terapeutica precoce si sustinuta , precum si profilaxia infectarii cu germeni redutabili este de o importanta covarsitoare. Acest deziderat poate fi indeplinit prin diagnostic precoce, urmat de monitorizare specifica , cu profilaxia infectiilor respiratorii, instituirea precoce a antibioterapiei si fizioterapie adecvata

MANAGEMENT OF PULMONARY STATUS IN CYSTIC FIBROSIS: UP-TO-DATE

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Background: Cystic fibrosis is the most common monogenic disease characterized by a marked clinical polymorphism, whose evolution is dictated by pulmonary disease, which is the main element in the prognosis of patients with cystic fibrosis

(FC). Bacterial infections are responsible for most of exacerbations in patients with CF, having frequently a polymicrobial etiology and resistant strains. Germs "traditional" associates FC are: *Pseudomonas aeruginosa, Staphylococcus aureus, Haemophilus influenzae, Burkholderia cepacia* complex while, *Stenotrophomonas maltophilia, Achromobacter xylosoxidans* are newer incriminated. Pulmonary status is influenced by the number and severity of exacerbations and disease progression is marked by infectious exacerbatins, leading to progressive loss of lung function and chronic respiratory failure. Acute respiratory functional and organic exacerbation lead to loss of lung parenchyma, first infection with Pseudomonas is frequently followed by colonization. The preservation of lung function is the main goal of therapy in CF because once lost, it cannot be recuperated. Antibiotic therapy should be instituted early in the appropriate dose and time according to the current consensus in conjunction with other therapeutic methods specific to physiotherapy.
In conclusion, early therapeutic intervention and support, and prevention of infection by germs redoubtable is of dominant importance. This can be achieved by early diagnosis followed by specific prophylactic monitoring of respiratory infection, early institution of appropriate antibiotic therapy and physiotherapy.

119. ADOLESCENTA CU HEMOPTIZIE, SFORAIT SI OBEZITATE!

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Introducere: Hemoptizia este unul din semnele alarmante in patologia respiratorie pediatrica, ridicand diverse suspiciuni etiologice. Hipertensiunea pulmonara de cauza non-cardiaca este rar intalnita la copil, spectrul etiologic incluzand pneumopatiilecronice, tulburarilede somn si obezitatea. Matreial si metode: Lucrarea prezinta cazul unei adolescente obeze de 13 ani , care este adresata clinicii cu istoric de hemoptizii si suspiciunea unei pneumopatii cronice obstructive. Anamnestic patologia pulmonara constand in tuse productiva si sputa hemoptoica a debutat cu 3 luni anterior internarii. Pacienta se afla in evidenta cu panhipopituitarism, primind tratament de substitutie hormonala. Investigatiile releva un sindrom de hipertensiune pulmonara, fara a se evidentia o patologie pulmonara care sa explice aceasta modificare. Polisomnografia evidentiaza o forma moderata de apnee de somn obstructiva. Remisia hipertensiunii pulmonare a fost semnificativa dupa instituirea diureticelor , inainte de tratamentul apneei de somn. Concluzie: Tulburarile de somn pot duce la hipertensiune pulmonara si la copil, mai ales in prezenta unor factori favorizanti/ declansatori ca obezitatea, tratamentul substitutiv hormonal.

ADOLESCENT GIRL WITH HEMOPTYSIS, SNORING AND OBESITY!

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Introduction: Hemoptysis is one of the signs alarming in pediatric respiratory pathology, raising suspicions various etiological. Pulmonary hypertension of non-cardiac cause is rare in children, including pneumopathies etiologic spectrum of chronic sleep disorders and obesity. Material and Methods: We report the case of an obese teenager of 13 years, which is addressed to the clinic with a history of hemoptysis and chronic obstructive suspicion of pneumopathies. Anamnesis lung pathology and productive cough consisting hemoptysis started with 3 months before admission. The patient is out with panhipopituitarism, receiving hormone replacement therapy. Investigations revealed pulmonary hypertension syndrome, without showing pulmonary pathology explaining this change. Polysomnography shows a moderate form of obstructive sleep apnea. Remission was significant pulmonary hypertension after setting diuretics prior to treatment of sleep apnea. Conclusion: Sleep disorders can lead to pulmonary hypertension and in children, especially in the presence of favorable factors / triggers obesity, hormone replacement therapy.

120. EFECTE POZITIVE ALE CORTICOSTEROIZILOR INHALANTORI IN TRATAMENTUL ASTMULUI PEDIATRIC

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Datorita schimbarilor climatice si antropogene din ultimii ani in Romania a crescut considerabil prevalenta astmului la copii. Nivelurile crescute de poluanti viabili (polen) si ne-viabili din atmosfera sunt asociate cu frecventa ridicata a astmului pediatric. De asemenea poluarea din spatiile inchise (fum de tigara) si conditiile precare de viata (mucegaiuri,etc.) amplifica incidenta astmului pediatric. Ca boala inflamatorie cronica a cailor aeriene astmul implica un model caracteristic de infiltrare a cailor aeriene cu limfocite, eozinofile, mastocite, depunere subepiteliala de colagen, cu hipertrofie si hiperplazie a musculaturii netede si de celule calciforme si glande submucoase. Deoarece consecintele pot fi mai severe la copii incluzand

simptome episodice sau persistente, hiperreactivitate bronsică, crize de bronhoconstrictie, controlul efectiv al astmului pediatric este o sarcină imperativ necesara. Simptomele astmului pediatric pot fi gestionate prin evitarea stimulilor de mediu, monitorizarea progresului bolii, folosind medicamente controler, si/sau utilizarea de medicamente de interventie rapidă. Lucrarea a evaluat eficienta terapiei astmului (inhibitori LT, corticosteroizi CSI, agonisti, sau o combinatie din acestea) aplicata unui grup de 53 de copii astmatici din mediul urban / rural Rosiori de Vede în perioada anului 2012. La sfârsitul studiului a fost redus numărul crizelor de astm bronsic, fiind imbunatatiti semnificativ parametrii respiratori functionali (VEMS si MEF25%, pulsoximetrie) si cei biologici (hemoleucograma, VSH, fibrinogen, eozinofile sangvine, numărul total de IgE). Prin controlul simptomelor de astm, imbunatatirea functiei pulmonare, prevenirea si reducerea exacerbarilor astmatice, s-au demonstrat efectele pozitive ale corticosteroizilor CSI administrate la majoritatea pacientilor copii si adolescenti cu astm, indiferent de vârstă sau severitatea astrmului.

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121 .CONSIDERATII CLINICO-EVOLUTIVE ASUPRA UNUI CAZ DE ASPERGILOZA LA COPIL

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Aspergiloza face parte dintr-un grup de boli determinate de microorganisme din genul Aspergillus. Majoritatea cazurilor pediatrice sunt cauzate de A. fumigatus. Astmul atopic poate fi precipitat de inhalarea sporilor specifici, ceea ce duce la un raspuns mediat Ig E si bronhospasm. Aspergiloza bronhopulmonara alergica se poate asocia la 7-10% din pacientii cu astm corticodependent.
Autorii prezinta cazul unui copil diagnosticat cu astm bronsic de la varsta de sugar, cu evolutie ondulanta. Sub tratament controller se adauga eozinofilie cu valori progresiv crescande, si in final un sindrom “hiper IgE” (valori > 4000UI). Sindromul inflamator este intens pozitiv. Se infirma orice alta etiologie infectioasa autoimuna sau neoplazica si se confirma aspergiloza bronhopulmonara alergica.
Cuvinte cheie: aspergiloza, IgE, copil

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122. BRONCHOPUMONARY ASPERGILLOSIS IN A CHILD – CASE PRESENTATION

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Aspergillosis refers to a group of diseases caused by monomorphic, mycelial fungi of the genus Aspergillus. Most aspergillosis in children is caused by A. fumigatus. Atopic asthma may be precipitated by inhalation of Aspergillus spores, which triggers an Ig E- mediated response and bronchospasm.
Allergic bronchopulmonary aspergillosis complicates chronic pulmonary disease in approximately 7–10% of patients with corticosteroid-dependent asthma.
The authors are presenting the case of a child diagnosed with asthma as infant with oscillating evolution. Under controller treatment, eosinophilia with progressively increasing values and finally a "hyper IgE" syndrome (over 4000 IU/ml) are added. Inflammatory syndrome is strongly and persistently positive. Any other infectious, autoimmune or neoplastic etiology are excluded and allergic bronchopulmonary aspergillosis is confirmed (specific IgE >4000 IU/ml).
Key words: Aspergillosis, IgE, children

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123. INFECTIA CU PSEUDOMONAS AERUGINOSA LA COPIII CU FIBROZA CHISTICA

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Introducere

Infectia pulmonara cronica cu Pseudomonas aeruginosa este dificil de eradicat si are un prognostic nefast in evolutia pe termen scurt si lung la pacientii cu mucoviscidoza. Este asociata cu o morbiditate semnificativa, calitate a vietii redusa, si în cele din urma cu o mortalitate crescuta la pacientii cu fibroza chistica (FC).

Material si metode

Infectia cronica cu P. aeruginosa a fost investigata la copii diagnosticati cu FC in Clinica II Pediatrie - Spitalul Clinic de Urgenta pentru Copii "Sf Maria" Iasi. Aspiratul hipofaringian a fost recoltat de la 45 de pacienti in timpul anului 2014, inainte de initierea oricarui tratament antibiotic.

Rezultate si discutii

Din cei 45 de pacienti testati P. aeruginosa a fost izolat in aspiratele hipofaringiene la 10 pacienti. Factorii de risc pentru infectie au fost: varsta la diagnostic, severitatea bolii si frecventa exacerbarilor, statusul nutritional precar, necomplianta la tratament, deficitele imune, mersul in comunitate si statutul socio-economic si educational precar al familiei. Infectii concomitente cu S. aureus, H. influenzae si alti germeni au fost regasite la 8 pacienti. Toti cei 10 pacienti au primit tratament antibiotic specific, probele colectate de la trei dintre acestea fiind negative dupa 2 cicluri de tobramicina in nebulizari.

Concluzii

Diagnosticul corect si masurile de tratament precoce pentru copiii infectati cronic cu P. aeruginosa pot influenta semnificativ istoria naturala a bolii. Depistarea precoce ofera o sansa pentru tratament si o mai buna calitate a vietii.

PSEUDOMONAS AERUGINOSA INFECTION IN CHILDREN WITH CYSTIC FIBROSIS

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Introduction

Chronic pulmonary infection caused by Pseudomonas aeruginosa is difficult to eradicate and it has an important influence regarding short term outcome and further evolution of the disease.This infection is associated with significant morbidity, reduced quality of life, and, finally, with increased mortality in patients with CF.

Material and methods

P. aeruginosa infection was investigated in children diagnosed with CF in the 2nd Pediatrics Clinic - Emergency Clinical Hospital for Children "Sf. Maria" in Iasi. Hypopharyngeal aspirate samples were collected from 45 patients during 2014. The collecting samples was performed before initiating any antibiotic treatment.

Results and discussions

Out of 45 patients tested, P. aeruginosa strains were detected in hypopharyngeal aspirates of 10 patients. Risk factors for infection were: age at diagnosis, disease severity and frequency of exacerbations, poor nutritional status, poor treatment compliance, immune deficits, attending community, socio-economic and educational status of the family. Concomitant infections with S. aureus, H. influenzae and other germs were found in 8 patients. All 10 patients received specific antibiotic therapy, the samples collected from three of those being negative after 2 courses of nebulised tobramycin.

Conclusions

Facilities and methods used to care for young children with CF can significantly influence their likelihood of acquiring P. aeruginosa in the respiratory tract and future evolution of the disease. Early detection provides a chance for treatment and a good quality of life.

TINERI CERCETATORI/REZIDENTI / YOUNG RESEARCHS

124. TROMBOZA VENOASA CEREBRALA : STUDIU RETROSPECTIV REALIZAT PE O PERIOADA DE 5 ANI

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Introducere : Tromboza venoasa cerebrala (TCV) este o conditie rara, cu o mare varietate de cauze. Pana la 35% din cazuri, nu au o cauza stabilita. Am realizat un studiu retrospectiv, pe o serie de pacienti cu TVC, internati in Spitalul Clinic de Urgenta pentru Copii "M. Sklodowska Curie", pe o perioada de 5 ani (2010-2014).

Metode : 25 de pacienti (20 baieti si 5 fete), cu varste incepand de la 1 zi de viata pana la 16 ani (varsta medie 3 ani), diagnosticati cu tromboza venoasa cerebrala cu ajutorul ecografiei Doppler, CT si IRM.

Rezultate : Au fost identificate 3 cazuri de trombofilie – doua cazuri cu deficit de proteina C si un caz cu deficit de proteina S. Predominant, 15 cazuri la pacienti diagnosticati cu otomastoidita, 2 pacienti cu sindrom nefrotic, 2 pacienti cu sepsis si 3 pacienti cu TVC idiopatica.

Concluzii : In urma studiului efectuat, se observa o tendinta de crestere a numarului de cazuri de TVC, in ultimii ani. Recunoasterea precoce a simptomatologiei si instituirea terapiei adecvate, duce la o evolutie favorabila, recuperare cat mai completa si poate reduce mortalitatea si morbiditatea.

CEREBRAL VENOUS THROMBOSIS : RETROSPECTIVE STUDY COMPLETED ON A 5 YEAR PERIOD

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Introduction : Cerebral venous thrombosis (CVT) is a rare condition, with a wide variety of causes. Up to 35% of cases have undetermined cause. We performed a retrospective study on a series of patients with CVT, admitted to Emergency Hospital for Children "M. Sklodowska Curie ", for a period of five years (2010-2014).

Methods : 25 patients (20 boys and 5 girls), with ages ranging from 1 day of life up to 16 years (mean age 3 years), diagnosed with cerebral venous thrombosis using Doppler ultrasound, CT and MRI.

Results : We identified 3 cases of thrombophilia - two cases with protein C deficiency and one case of protein S deficiency, 15 patients diagnosed with otomastoiditis, 2 patients with nephrotic syndrome, 2 patients with sepsis and 3 patients with idiopathic TVC.

Conclusions : In the study conducted, we have seen an increasing trend in the number of diagnosed cases of CVT in recent years. Early recognition of symptoms and appropriate treatment setting leads to a favorable trend, more complete recovery and can reduce mortality and morbidity.

125. O CAUZA MECANICA DETERMINANTA A SINDROMULUI/SECVENTEI PIERRE-ROBIN

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Sindromul/secventa Pierre-Robin nu are o cauza exacta stabilita; exista mai multi factori incrimnati in producerea lui: mecanici, genetici, neurologici. Este extrem de important a stabili caza malformatiei pentru a putea acorda sfatul genetic parntilor in privinta sarcinilor ulterioare.

Va prezint cazul unui sugar in varsata de 3 luni internat in clinica noastra pentru hipotrofie ponderala. La examenul obiectiv efectuat se ridica suspiciunea de sindrom/secventa Pierre-Robin: prezenta dismorfism facial, cheilopalatoschizis, micrognatism, glosoptoza toate indicand spre acest sindrom. Asociat pacientul avea defect de sept ventricular, persistenta de canal arterial, pete vasculare, hipospadias, testicoli necoborati, hernie inghinala bilateral. Aceste asocieri sugerau prezenta unui sindrom genetic. S-a efectuat cariotip care a fost normal, stabilindu-se astfel ca e vorba de secventa si nu de sindromul Pierre-Robin.

Din moment ce cauza genetic a fost inlaturata ramanea intrebarea inca valabila in dreptul celorlalte mecanisme etiopatogenetice. La anamneza amanuntita am descoperit ca mama a prezentat in antecedente o colecistectomie urmata de o eventratie care a fost operata si s-a tratat printr-o plasa a peretelui abdominal. Sarcina a decurs fiziologic pana la varsta de 36 saptamani cand a fost detectat ecografic un retard de crestere intrauterina a fatului motiv pentru care s-a efectuat sectiunea cezariana.

In concluzie am interpretat cazul ca fiind o secventa Pierre-Robin datorata unui factor mecanic, reprezentat de plasa abdominala materna care a determinat atat retardul de crestere intrauterin cat si malformatiile faciale.

CASE REPORT: A MECHANIC CAUSE FOR PIERRE-ROBIN SYNDROME/SEQUENCE

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Syndrome / sequence Pierre-Robin has not an exact cause established; there are several factors involved in producing it: mechanical, genetic, neurological. It is extremely important to establish the cause of malformation in order to offer genetic counseling for parents about future pregnancy.

I present the case of a 3 months infant admitted to our clinic for low weight. Performing the physical examination a suspicion was risen: Pierre-Robin syndrome / sequence. The infant presented facial dysmorphism, cleft palate aand cleft lip, micrognathism, glosoptosis, all pointing to Pierre-Robin syndrome / sequence. Associated, the patient presented ventricular septal defect, patent ductus arteriosus, vascular patches, hypospadias, undescended testicles, bilateral inguinal hernia. These associations suggest the presence of a genetic syndrome. It was performed a karyotype which was normal, thus establishing it is the Pierre-Robin sequence not the syndrome.

Since genetic cause has been removed, the question still stands in right about the other ethiopathogenetic mechanisms. Performing a detailed history I discovered that the patient’s mother has experienced a cholecystectomy followed by a eventration who underwent surgery and was treated by placing an abdominal wall prosthetic mesh. Pregnancy went up

physiological till 36 weeks of gestation when an ultrasound detected fetal intrauterine growth retardation, so a cesarean section was performed.
In conclusion we interpreted the case as Pierre-Robin sequence due to a mechanical factor represented by the maternal abdominal mesh which caused both intrauterine growth retardation and facial malformations.

VARIA

126. REDUCTION OF PAIN IN VENIPUNCTURE IN CHILDREN

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INTRODUCTION:The entrance to hospital for the child is, at times, a source of fear often linked to venipuncture, practice frequently used for diagnostic and therapeutic. The nurse who approaches the child must be able to handle the problem in order to deliver answers assistance reducing the emotional impact on the child.
OBJECTIVE: the study consisted of the comparison between two different methods to reduce pain correlated to venipuncture, concluding which of them is better in children between the age of 4 and 12.
MATERIAL AND METHODS: we used two methods, active distraction and cryoanesthesia, because supported by scientific literature. The study included 70 children, random divided in two groups, both having a venipuncture: the first group was asked to inflate a balloon during the procedure, the second group received an ice-cube to apply for a minute on the skin before the venipuncture. We evaluated the pain's intensity using the Wong-Baker Faces Pain Scale (rating of perceived pain) and CHEOPS scale (behavioral rating scales).
RESULTS AND CONCLUSIONS: all children were in the same situation, the caregiver and a personal nursery care were allowed. The intensity of venipuncture's pain was different in children, depending on the method used to reduce it during the procedure. The active distraction with inflation of a balloon was found to have greater efficacy compare to crioanaesthesia. The first one is cheaper and very quick to apply,too.

127. FROM SIGN TO SYMPTOMS

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INTRODUCTION: the report will aim to joint the medicine interesting organic failure and medicine related to the psychic, the first one related to signs and the second one to the symptoms. "Heath is in the silence of the organs", wrote Rene Leriche in the early twentieth century: it is healthy if the body doesn't speak.
DISCUSSION: we're talking about Psycosomatic, the Medicine jointing symptoms and emotional factors , together involved in the Human body. "From sign to symptom", we often confuse their definition :the sign is objective and it can be measurable , the symtom is subjective and related to the person referring it. "Somatoform disorders", placed in the Diagnostic and Statistical Manual of Mental Disorders (DSM), are physical diseases without a diagnostic evidence, because of the absence of a pathogenetic mechanism. Stress is the main trigger and it can be an useful reaction to different situations, as already reported in studies of Seyle in 1936. Emotional stress is coded and processed, a reaction is following, the brain speaks through autonomic nervous system, made of mediators acting at central and peripheral receptors.
MATERIAL AND METHODS: we considered 300 children and adolescents between 6 to 16 age, evaluating the main psycosomatic disorders: digestive disorders (dyspepsia, vomit, abdominal pains), cardiovascular disorders (tachicardia, syncope), eating disorders (anorexia, bulimia), respiratory disorders (asthma, air hunger, cough), growth's disorders, other (paresthesias, hyperhidrosis, sleep disorders)

CONCLUSIONS: the somatic involvement is very frequent in children and adolescents. Family history and education play a key role. Children and adolescent showing their emotions are less at risk, compared to those who introject. The symptom is brought to medical attention when it can't longer be placed in a pattern of defence. "In puero homo", the pediatrician has a role of great responsability as a social function, as "advocate".

128. DIABETUL ZAHARAT TIP 2 SI INSULINOREZISTENRA LA COPII SI ADOLESCENTI: DE LA TEORIE LA PRACTICA

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Boala metabolica complexa, cu etiologie heterogena, caracterizata prin hiperglicemie cronica, in care predispozitia genetica se transforma in boala sub influenta factorilor de mediu, Diabetul Zaharat tip 2 se contureaza ca o noua problema in practica clinica pediatrica, incepand cu anii '90, cunoscand o tendinta de crestere a prevalentei in randul copiilor si adolescentilor paralel cu cresterea obezitatii pe plan mondial. Termenul "epidemic" este folosit frecvent pentru a descrie amenintarea acestei patologii complexe, dar constatarile practicii clinice nu sustin - deocamdata - aceasta presupunere, mai ales in tarile europene.
Sunt discutate componentele cheie in patogeneza Diabetului Zahara tip 2: insulinorezistenta si disfunctia celulei beta-pancreatice, ca si particularitatile de evolutie specifice varstei pediatrice: declinul mai rapid al functiei celulei beta-pancreatice si instalarea precoce a complicatiilor cronice micro- si macroangiopatice, aspecte care impun o abordare terapeutica intensiva inca de la inceputul bolii. Din multitudinea de medicamente disponibile pentru pacientii adulti cu diabet zaharat tip 2, doar metforminul si insulina au trecut, pana in prezent, testele de siguranta si eficacitate. Desi par usor de implementat, regulile unui stil de viata sanatos - alimentatie echilibrata si miscare – constituie o permanenta provocare, atat pentru pacient cat si pentru echipa medicala, fiind cheia terapeutica si mai ales de preventie a bolii si complicatiilor acesteia.

TYPE 2 DIABETES MELLITUS AND INSULIN RESISTANCE IN CHILDREN AND ADOLESCENTS: FROM THEORY TO CLINICAL PRACTICE

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A complex metabolic disease, characterized by chronic hyperglycemia, with a heterogeneous aetiology in which genetic predisposition is turned into disease under the influence of environmental factors, type 2 diabetes mellitus is emerging as a new problem in pediatric clinical practice since the 90s, with an increasing trend in prevalence among children and adolescents in parallel with the rise in obesity worldwide. The term "epidemic" is often used to describe the threat of this complex disease, but clinical practice do not support - for now - this assumption, especially in the European countries.
Here are discussed the key components in the pathogenesis of type 2 diabetes: insulin resistance and dysfunction of pancreatic beta cell, as pediatric age-specific peculiarities of evolution: cell function decline faster beta-pancreatic and early installation of chronic micro- and macroangiopatic complications in younger onset, issues requiring intensive therapeutic approach since the beginning of the disease. Of the many drugs available for adult patients with type 2 diabetes, only metformin and insulin past, so far, tests of safety and efficacy. Although they seem easy to implement, the rules of a healthy lifestyle - eating a balanced diet and exercise - are a permanent challenge for both the patient and medical team, but this is a the key of therapeutics and prevention of the disease and its complications.

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