

POLYDACTYLY OF THE HAND AND FOOT CASE REPORT

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Abstract

Congenital malformations of the limbs are among the most frequent congenital anomalies found in humans, and they preferentially affect the distal part, the hand or foot.

They exhibit a wide spectrum of phenotypic manifestations and may occur as an isolated malformation and as part of a syndrome.

We report a case of both hand and foot polydactyly in a patient from a family were for three generations all males have been affected by anomalies regarding the number of digits on hands or feet.

Key words: congenital deformities of the hands, polydactyly, limb malformations.

Introduction

Polydactyly is one of the most common congenital deformities of the hands. It can occur as an isolated disorder, in association with other malformations of the hands or feet, or as part of a syndrome. It can occur sporadically but it can also be inherited with a mainly autosomal dominant inheritance. (Karaaslan 2003)

Genetic developmental malformations on the limbs, in man, in many instances do not interfere with reproductive fitness, yet they are likely to be recognized and reported by the physician.

A common and conspicuous congenital hand anomaly, polydactyly commonly involves only the hand or the foot. Polydactyly involving both hands and feet is rare. (Hosalkar 1999)

Case report

Our case is a female, the first born child of healthy non consanguineous parents aged 25 and 28 years. There was no previous history of other pregnancies or abortions for the mother. The patient was born at 40 weeks of gestation with a normal body weight (3100g) and both head circumferences and body length within normal range.

Family history was positive for hand and foot malformations. For three generations the males of the family were affected by both hand and foot polydactyly or just by isolated hand or foot malformations. Both the father of the child and his brother have hand polydactyly but no other associated malformations. A familial pedigree analysis suggested that polydactyly was inherited as an autosomal dominant trait in the family.

Postnatal examination revealed a hand and foot polydactyly with 6 finger bilateral hands and 7 toes at both legs. Other than these findings there were no other malformations or conditions noted after intensive investigation. (Figure 1. A, B).

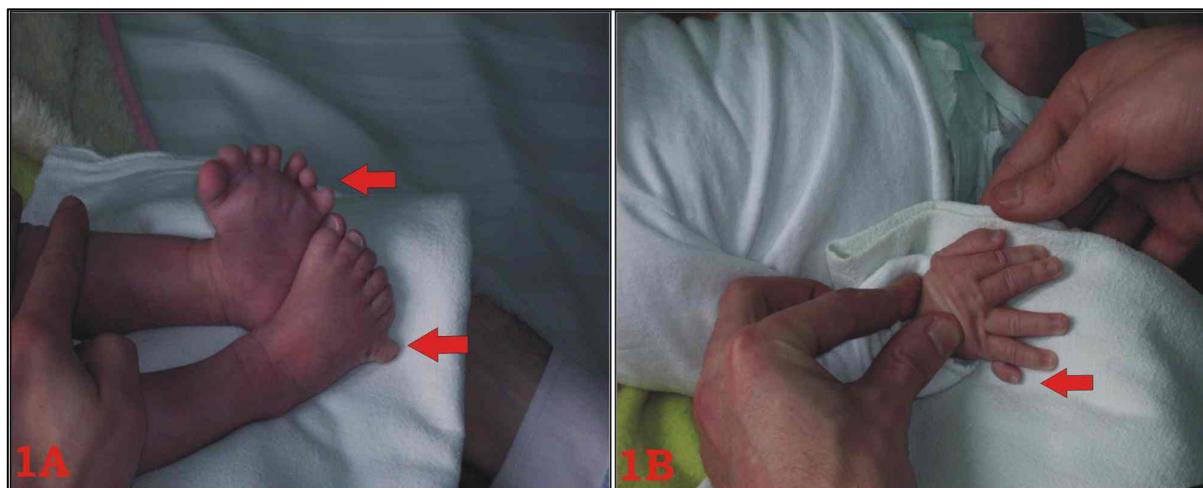


Figure 1. Hand (1B) and Foot polydactyly (1A).

Postnatal X-ray of the patient's hands and feet was performed in order to determine which surgical approach should be considered.

At the level of his hands, the 6th finger was composed of two phalange segments of cartilaginous origin without any modifications to be mentioned regarding carps and meta-carps. The postaxial digits in the hands were floating, with no palpable bones or active movements. Further clinco-radiological examination revealed no other congenital anomaly.

Discussions

The patient described in this report is one of the 6 cases of both hand and feet polydactyly, that we encountered in a retrospective study from 1995 to 2005 conducted at the Children's Hospital "Louis Turcanu", Department of Pediatric Surgery in Timisoara. Out of 64 cases of polydactyly, in 41 cases the hands were affected (13 left, 19 right, 9 both hands), in 17 cases the feet were malformed (4 left, 7 right, 6 both feet) and only in 6 cases both hands and feet were affected.

With regards to the surgical correction of the polydactyly, is almost always indicated, not only for cosmetic improvement but also for better function. Surgical reconstruction generally is performed between 18 months to 5 years.

The presence of extra digits is the most common limb deformity of the human hand and is the consequence of disturbances in the normal program of limb development. However, despite the extensive use of the developing limb as a classical developmental model, the cellular and genetic mechanisms that control the number and identity of the digits are not completely understood. (Talamillo and Bastida 2005).

There are several growth factors that can modify in certain conditions the outcome of the limb development. Fibroblastic growth factors like FGF-2, FGF-4 and FGF-8 are just some of the elements that cause the mesodermal cells to proliferate and expand outward in different directions. FGF8 is unique not only in its expression pattern, but also because it is the only such FGF gene that causes limb skeletal abnormalities, like polydactyly, when individually inactivated. (Lu 2006)

Lu et al. (2006), reported that the increase in FGF signaling that occurs when the FGF4 gain-of-function allele is activated in a wild-type limb bud causes formation of a supernumerary posterior digit, postaxial polydactyly, as well as cutaneous syndactyly between all the digits.

In recent years, increasing knowledge of the molecular basis of embryonic development has significantly enhanced our understanding of congenital limb malformations.

References:

1. Lu P, Minowada G, Martin GR. (2006). Increasing Fgf4 expression in the mouse limb bud causes polysyndactyly and rescues the skeletal defects that result from loss of Fgf8 function. *Development*. Jan; 133(1): 33-42
2. Schwabe GC, Mundlos S. (2004). Genetics of congenital hand anomalies. *Handchir Mikrochir Plast Chir*. Apr-Jun; 36(2-3): 85-97.
3. Talamillo A, Bastida MF, Fernandez-Teran M, Ros MA. (2005). The developing limb and the control of the number of digits. *Clin Genet*. Feb; 67(2):143-53
4. Karaaslan O, Tiftikcioglu YO, Aksoy HM, Kocer U (2003) Sporadic familial polydactyly. *Genet Couns*. 2003;14(4):401-5.
5. Hosalkar HS, Shah H, Gujar P, Kulkarni AD (1999) Crossed polydactyly. *J.P.G.M.* 99 ; 45 ; 3 ; 90-2

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