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Case Report Open Access

A Case of Complete Cutaneous Syndactyly of the Toes with Non-Syndromic Phenotype

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Abstract

Cutaneous syndactyly is a malformed condition in which fingers are joined together. It is one of the most common hereditary limb malformations depicting the fusion of certain fingers and/or toes, and can involve the bones or just the skin. The type affecting the feet by the fusion of two or more toes may occur as an isolated entity with non-syndromic phenotype or a component of many genetic syndromes: principally Apert, Carpenter, and Smith-Lemli-Opitz. We present the case of a healthy 2-year-old boy with a complete cutaneous syndactyly, bilateral and near symmetrical in both feet, which appears to represent a non-syndromic phenotype that is unclassified and previously undescribed.

Keywords: Cutaneous syndactyly; Syndactyly with non-syndromic phenotype

Introduction

Syndactyly of the foot is a common congenital abnormality in which there is persistence of webbing between adjacent toes. It may involve fusion of the soft tissues with or without bony fusion; cutaneous syndactyly of the toes is soft-tissue continuity in the anterior/posterior axis between adjacent foot digits that involves at least half of the length of one of the two involved digits [1]. Cases lacking other combined limb anomalies and without affected first-degree relatives are rare [2]. The main cause is the failure of differentiation between adjacent digits by the absence of apoptosis in the interdigital mesenchyme during the second month of gestation. The most common site is between the second and the third toes; the condition is frequently bilateral. Inheritance is thought to be autosomal dominant with variable penetrance and expressivity, with male predominance in half of the cases; the other cases correspond to a genetic sporadic appearance [3]. Foot syndactyly can be an isolated finding or seen with other anomalies as a feature in several syndromes like Apert, Carpenter, and Smith-Lemli-Opitz.

Case Report

We present a healthy 2-year-old boy (Figure 1) of first gestation and full-term pregnancy from healthy young parents in a non-consanguineous marriage. There was no maternal smoking or exposure to toxics, drugs, or medicines during the pregnancy, and the childbirth was without complications. At birth, newborn was discovered to have complete cutaneous syndactyly in both feet. There was negative familial history of relatives with this condition. The physical examination without associated malformations showed: simple cutaneous syndactyly involving only soft tissues with no bony fusion, complete because the entire proximodistal length of the digits was affected (Figure 2); all the adjacent toes from the first to the fifth toes were fused to the finger tip of the distal phalanx as a multiple type. This was bilateral and very symmetrical. The great toe and second toe shared neighbouring nails that were almost fused but there was not actually a common nail (synonychia) (Figure 3); they had distinct nail plates with a trough separating them. There was a slight wrinkle between the fourth and fifth digits with light overriding of the fifth toe. The plantar region showed a fusion of the toes with almost no interdigital separation (Figure 4). The results of X-ray examination of the both feet showed no bone affection (Figure 5). The laboratorial blood and urine test were normal, rule out an inborn error of metabolism. All the measures of skull, using the bony reference points: nasion, bregma, lambda and cephalic perimeter biauricular and antero-posterior distances, expressed median cranial-facial index with proportional growth rate and shape at his sex and age. The genetic study shows normal male karyotype at 550 band level: 46, XY with standard chromosome morphology, no abnormalities; and there were no craniofacial abnormalities.

Figure 1: Normal face without craniofacial anomalies

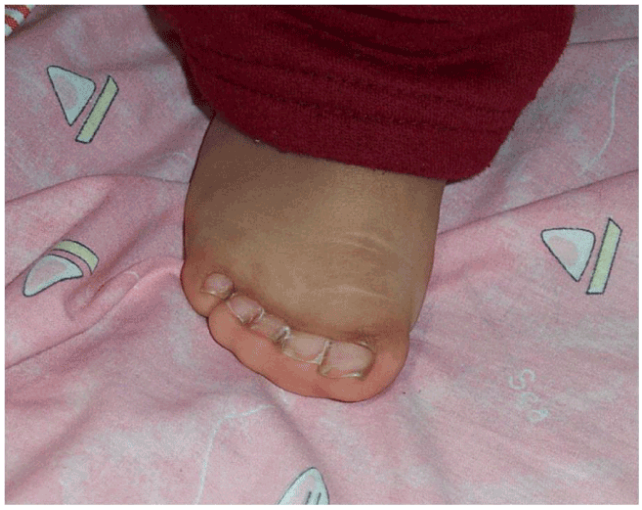

Figure 2: Right foot: all the adjacent toes are fused along the entire length.

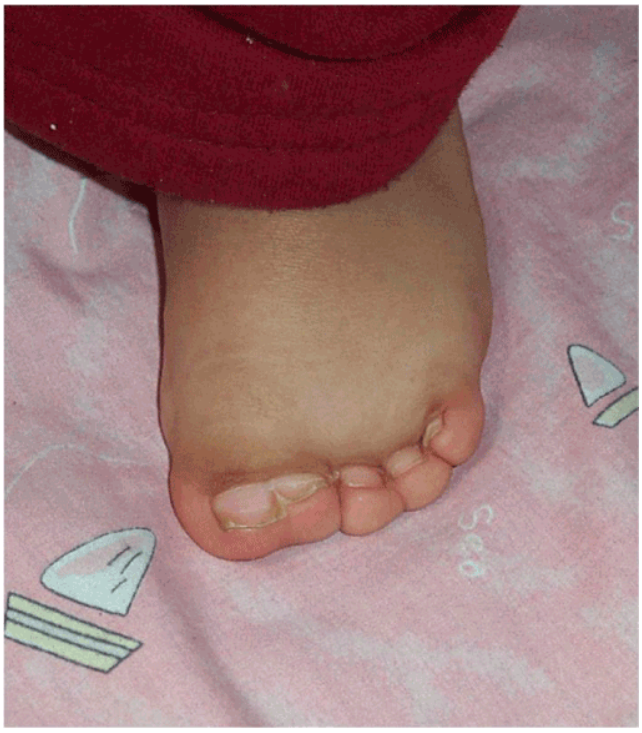

Figure 3: Left foot: the great toe and second toe share almost-fused nails.

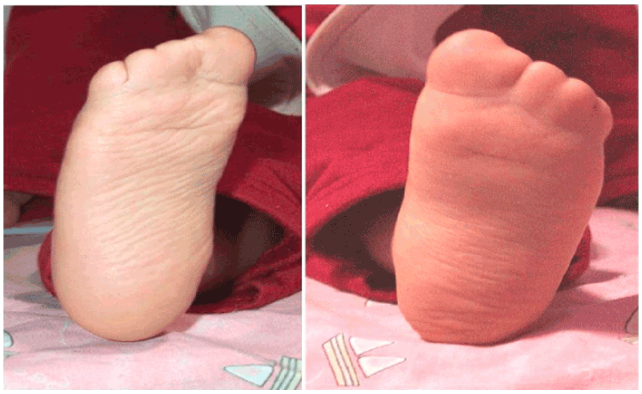

Figure 4: Plantar view of the feet shows complete cutaneous syndactyly.

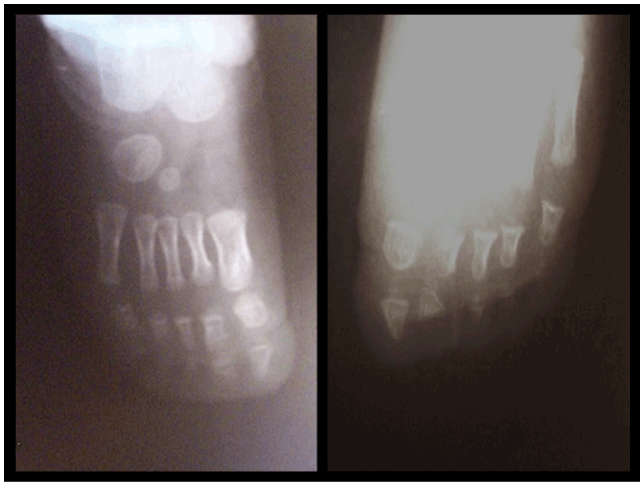

Figure 5: Feet x-ray images show no fusion of the bones.

Discussion

Syndactyly of the foot is a common congenital abnormality in which there is persistence of webbing between adjacent toe, can also occur along with other birth defects involving the skull, face, and bones. It is caused by the failure in separation of digital rays; the lack of digital differentiation during the embryonic period [4]; and if apoptosis or programmed cell death during gestation is absent or incomplete. Syndactyly of the foot affects around 1/2000 people [5]. More common causes include Down syndrome and hereditary syndactyly, for some children, syndactyly is only one feature of a more complex genetic condition or other syndrome like Apert, Carpenter, Pfeifeer or Smith-Lemli-Opitz syndrome.

Non-syndromic is inherited as an autosomal dominant trait, although the more severe and sub appear to have autosomal recessive inheritance. The patient did not correspond to any of the nine previously reported non-syndromicsyndactyly phenotypes [6] but resemble type IV of Temtamy, a very rare type with only four reports in the literature. Haas was the first to describe it as affecting the fingers of both hands [7]. No cases of the feet have been reported and its gene localisation has not yet been assigned. However, novel mutations in the homeobox d13 (HOXD13) gene may be the cause of the different phenotype like this case of of all toes in both feet without characteristic craniofacial anomalies or any syndromic phenotype [8]. This case may correspond to a new, previously unrecognised sporadic mutation. Further observation of new cases will be required to determine the gen implicated [9]. The soft-tissue syndactyly in the feet of this patient caused neither disability nor functional loss and did not impair the ability to perform any activity, including walking or running. Cases of syndactyly usually do not require repair; the aim of web reconstruction for syndactyly of the foot is purely cosmetic [10], to prevent the associated significant psychological morbidity and allow the patient to gain social acceptance [11].

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