Case Report

A rare case of ectrodactyly in a child in India

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ABSTRACT

Ectrodactyly, termed split-hand/split-foot malformation (SHFM), is a rare genetic condition characterized by defects of the central elements of the autopod (hand/foot). Clinical presentation is with the absence of one or more median rays or digits creating cone-shaped clefts of the hands and/or feet. The present case of severe bilateral SHFM was presented in an 8-year-old girl in India. This case of SHFM involves the complete absence of the central rays of the autopod in which each of the hands and feet is divided into two parts by a cone-shaped cleft tapering proximally, resembling a "lobster claw." SHFM is often associated with other limb anomalies, including monodactyly, syndactyly and aplasia, and/or hypoplasia of the phalanges, metacarpals, and metatarsals. Most cases are sporadic; however, familial forms do exist with predominantly autosomal dominant inheritance. This case is an example of the non-syndromic form of SHFM expressed with isolated involvement of the limbs, while the syndromic form is associated with anomalies such as intellectual disability, ectodermal and craniofacial findings, and hearing loss. Non-syndromic isolated ectrodactyly does not usually require surgical intervention. We recommended against surgical reconstruction due to lack of evidence of functional disability.

Keywords: *Congenital limb deformities, ectrodactyly, split-hand/split-foot malformation*

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Introduction

Split-hand/foot malformation (SHFM) is a primary structural rare abnormality of the limbs. SHFMor ectrodactyly is characterized by the absence of one or more median rays or digits creating cone-shaped clefts of the hands and/or feet. It is often associated with other limb anomalies, including monodactyly, syndactyly and aplasia, and/or hypoplasia of the phalanges, metacarpals, and metatarsals.

We report herein a case ofsevere SHFM involving all four limbs.

Case Report

An 8-year-old girl presented withsevere bilateral split-hand and foot malformations since birth [Figure 1]. Physical examination revealed absence of multiple metatarsals and phalanges with a cone-shaped cleft tapering proximally in both the feet and in both the hands. There were no other medical conditions or dysmorphic features. The girl was an only child, born out of a non-consanguineous marriage. Herparents are non-syndromic with no family history of limb malformations. The parents brought their daughter to the medical center for consultation regarding options for treating what they perceived as a cosmetic deformity. Examination showed no functional disability, thus surgery was not recommended.

Discussion

Ectrodactyly, also termedsplit-hand/split-foot malformation, is a rare genetic condition characterized by defects of the central elements of the autopod (hand/foot). It has a prevalence of 1:10,000-1:90,000 worldwide.

The presented case of severe bilateral SHFM involves the complete absence of the central rays of the autopod in which each of the hands and feet is divided into 2 parts by a cone-shaped cleft tapering proximally, resembling a "lobster claw".[1] The severity may vary between patients as well as between different limbs of the same individual.

Most cases of SHFM are sporadic as in this case report; however, familial forms do exist with predominantly autosomal dominant inheritance. Autosomal-recessive and X-linked forms are rare while some cases of SHFM are due to chromosome deletions and duplications.[2]

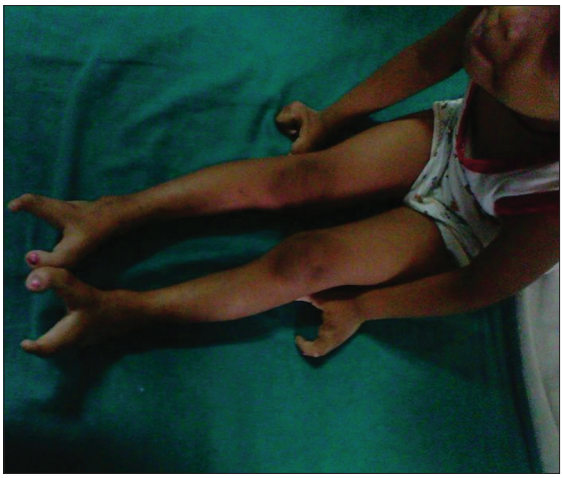

Figure 1: Image of girl presented with severe bilateral split-hand and foot malformations

The main pathogenic mechanism for ectrodactyly development is due to a failure to maintain median apical ectodermal ridge (AER) signaling, either through increased cell death, or through reduced cell proliferation. This AER activity defect does not occur in the very earliest stages of limb development, since that would result in more severe limb malformations that are not limited to the autopod.[2]

The syndromic form of SHFM expressed in some patients is associated with anomalies asintellectual disability in 33%, ectodermal and craniofacial findings in >35% and orofacial clefting and neurosensory hearing loss in >35%.[3]

The malformation, when non-syndromic can be expressed with isolated involvement of the limbs, as presented in this case. Usually cases of isolated ectrodactyly do not require surgical intervention. We also recommended against surgical reconstruction, which should be considered only in cases with functional disability. Most individuals adapt well and live normal lives with modest functional impairment of the hands. Thus, functional training and physical therapy are advised.

References

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