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Case Series

Isolated Ectrodactyly/Split Hand-Feet Malformation: About Two Cases

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Abstract

Ectrodactyly is a rare congenital malformation of the limbs, of variable severity, affecting more specifically the central rays of the autopod. Diagnosis is possible in antenatal by obstetrical ultrasound and radiological examinations are requested as part of the pre-therapeutic assessment in order to judge the possibility of a surgical intervention which may be indicated in severe forms in order to improve the functioning of the affected limbs.

Keywords: *Ectrodactyly; Lobster Claw; Congenital Malformation; Case Report*

Introduction

Ectrodactyly or SHFM is a rare congenital malformation whose incidence varies between 1 and 9/100,000 births, affecting the limbs and more specifically the central rays of the autopod, with variable severity [1,2]. We report two cases of bilateral ectrodactyly of the upper and lower limbs in two sisters with studies of the clinical characteristics of this malformation, its pathophysiology and management.

Observation 1

A 7-year-old girl from a full-term pregnancy with good psychomotor development who presented with deformed hands and feet since birth. Both hands had clefts with agenesis of the thumb, index, middle, and ring fingers on the right and the thumb, index, and middle fingers on the left (Figure 1A). Standard radiographs of the hands confirmed agenesis of the phalanges of the right 2nd, 3rd and 4th radii with total absence of the thumb and on the left the absence of the thumb with agenesis of the phalanges of the 2nd and 3rd fingers (Figure 1B). Concerning the feet, she presented on the right foot a total agenesis of the 2nd, 3rd and 4th radii realizing a "lobster claw" aspect also classified as type V in the classification of Blauth and Borisch. On the left foot, she had a monodactyl cleft with total agenesis of the 1st, 2nd, 3rd and 4th radii classified as type VI in the Blauth and Borisch classification (Figure 1C and 1D). A CT scan of both feet in the bone window was performed as part of the pre-therapeutic workup, showing the above malformations (Figure 3A and 3B).

Observation 2

An 11-year-old girl also presented with congenital deformity of the hands and feet. Both hands showed clefts and agenesis with total agenesis of the thumb and index finger on the right, agenesis of the phalanges of the middle finger as well as the middle and distal pha-

langes of the ring finger on the right, and absence of the thumb and phalanges of the fifth finger on the left (Figure 2A and 2B). The feet showed a bilateral monodactyly cleft with individualization of only the fifth metatarsal and the toe and total agenesis of the first, second, third and fourth radii, classified as type VI of the Blauth and Borisch classification (Figure 2C and 2D). In this case, a CT scan of both feet in the bone window was also performed as part of the pre-therapeutic workup, showing the same deformities (Figure 3C and 3D).

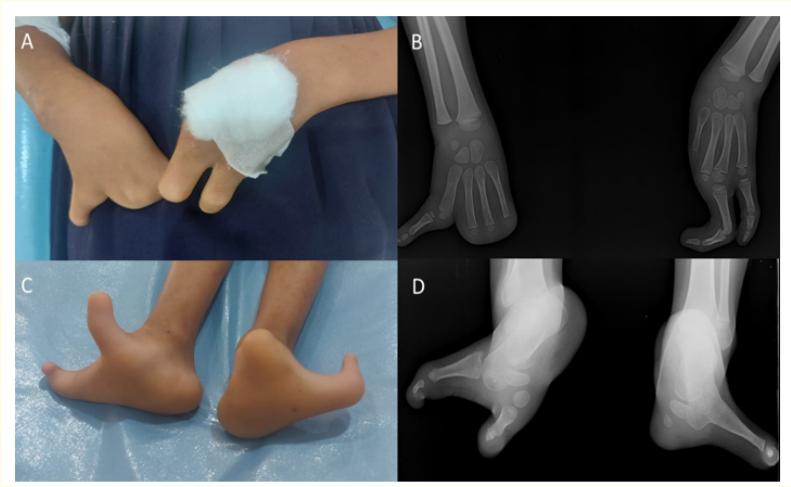

Figure 1: *Split hand/split feet showing syndactyly, median clefts of the hands and feet and aplasia/or hypoplasia of the phalanges, metacarpals and metatarsals.*

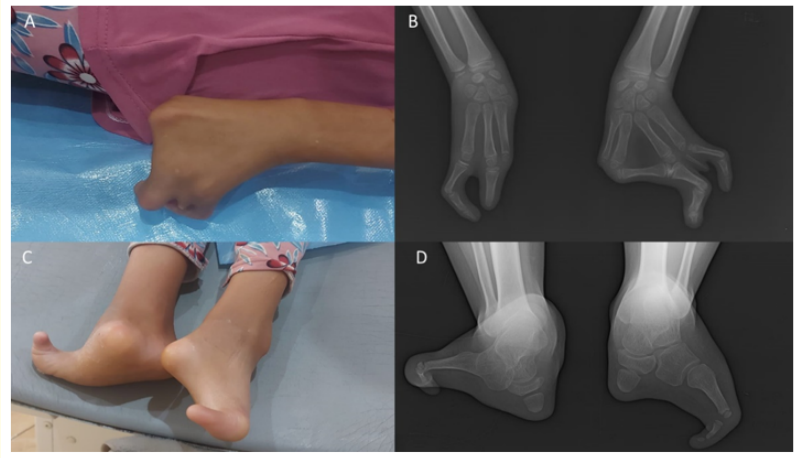

Figure 2: *Ectrodactyly of all four limbs.*

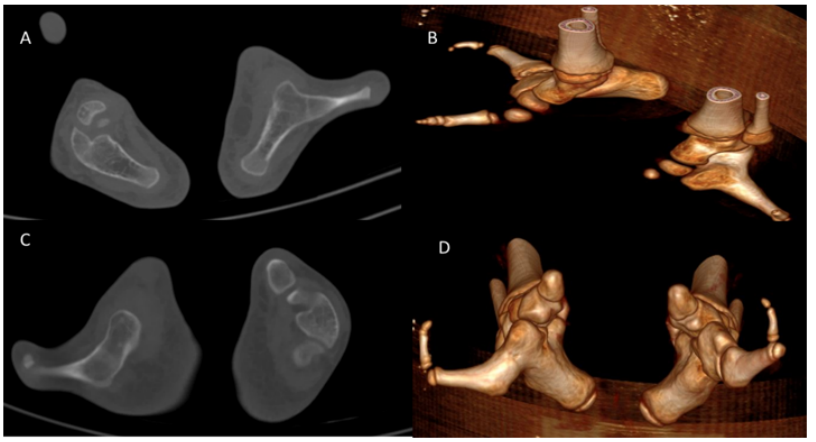

Figure 3: *CT images in bone window (A, C) and in 3D reconstruction (B, D), showing ectrodactyly of both feet.*

Discussion

SHFM/ectrodactyly is a rare congenital malformation of the upper and lower limbs affecting the extremities, corresponding to an absence of the central rays of the fingers with a cleft in the hands and feet often associated with syndactyly, oligodactyly and rarely with polydactyly realizing the characteristic "lobster claw" appearance [1,2]. Usually, it affects quarter limbs with a family history proving its genetic origin [3]. This is the case of the two sisters of our observation who presented malformations of the four limbs. Ectrodactyly can be an isolated limb disorder known as the non-syndromic form, or it can be associated with other malformations such as tibial aplasia, ptosis, mental retardation, craniofacial malformations and deafness [3,4]. Our cases belong to the non-syndromic type of SHFM due to the absence of associated anomalies. Genetically, seven chromosomal loci have been found in isolated SHFM: SHFM 1 to 6 and SHFM/SHFLD. The transmission depends on the chromosomal location: SHFM 3 and 4 are transmitted in an autosomal dominant mode, SHFM2 is X-linked recessive, SHFM1 follows, without certainty, an autosomal dominant transmission pattern with a high penetrance and very variable expressivity [4-6]. In our case, the inheritance pattern is probably autosomal recessive because only the two sisters were affected without any other family member.

Pathophysiologically, the formation of developing membrane buds is mediated by signaling molecules produced by groups of specialized cells. These constitute the zone of progression (PZ), the zone of polarizing activity (PZA) and the apical ectodermal ridge (AER). A defect in the maintenance of the AER leads to the development of SHFM, which can also be caused by all environmental factors that affect the AER [7]. Clinically, this malformation is characterized by a syndactyly corresponding to a joining of two fingers or two toes together, with oligodactyly i.e. agenesis of one or more fingers or toes on one hand or foot. This results in a median split in the hands or feet with opposition of the fingers, creating a "lobster claw" appearance. Other anomalies may be observed, of varying severity, ranging from simple agenesis of a single central ray to monodactyly where only one finger is present [1,3,4].

The diagnosis of ectrodactyly is made at birth during the clinical examination. However, it is possible to detect these malformations during pregnancy by means of antenatal ultrasound [8].

Radiological examinations, including standard X-rays and bone window scans of the limbs, are requested as part of the pre-treatment workup in order to assess the possibility of surgical intervention.

No medical treatment is available for this type of malformation. However, a surgical reduction and a fitting are possible to improve the aesthetics and the walking in children whose life is impacted by this malformation [8]. In our case, it is a major form with a preserved functional potential.

Our patients were referred to a pediatric surgical service for possible follow-up.

Conclusion

The split hand/foot malformation known as ectrodactyly is a rare skeletal malformation of congenital origin, with an etiopathogeny that is still poorly elucidated, characterized by a great inter- and intra-individual variability. Its management must be multidisciplinary with recourse to surgical treatment in severe forms in order to improve the prognosis.

Conflict of Interest Statement

No conflict of interest.

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