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Monomelic Ulnar Dimelia Associated with Ipsilateral Polydactyly: An Extraordinary Anomaly

Monomelic Ulnar Dimelia Associated with Ipsilateral Polydactyly: An Extraordinary Anomaly
Radyoloji

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Özet

Ulnar dimeli (ayna deformitesi), polidaktili, radiyus yokluğu ve ulnar duplikasyon ile karakterize nadir bir konjenital bozukluktur. Bu yazıda, aynı taraflı polidaktilil ile birlikte olan unlar dimeli'li nadir bir olguyu sunuyoruz.

Anahtar kelimeler: *Ayna deformitesi, Polidaktili Ulnar dimeli Direkt radyografi*

Abstract

Ulnar dimelia (mirror deformity) is a rare congenital disorder that is characterized by the duplication of the ulna, absence of the radius and polydactyly. Herein, we described a rare case of ulnar dimelia associated with ipsilateral polydactyly.

Keywords: *Mirror deformity, Polydactyly Ulnar dimelia Plain radiograpy*

Introduction

Ulnar dimelia (mirror deformity) is a rare dysontogenetic deformity of the upper limb caused by an embryonic disturbance of the developing limb bud. It is characterized by two ulnae, the duplication of the ulnar halves of the carpus, metacarpals and phalanges, with the absence of the radius and thumb. It is commonly unilateral. This anomaly may be associated with shoulder dislocation, fibular dimelia, idiopathic scoliosis, cirrhosis, pyloric hypertrophy or a polycystic kidney. Approximately 72 cases have been reported since it was first described in 1587 [1-3]. We herein describe a rare case of ulnar dimelia associated with ipsilateral polydactyly.

Case Report

A 2-year-old girl presented with six digits on her right hand. She was born at term after an uneventful pregnancy. She was the first child. Both parents were healthy and did not have any limb abnormalities. Family history was unremarkable. Chromosomal analysis showed a normal 46, XX karyotype. All physical findings were normal except for the right upper limb, characterized by a right forearm that was shorter than the left forearm. Wrist movements were normal. Flexion of the elbow and supination/pronation of forearm were limited to 40 degrees and 10 degrees, respectively. There were no apparrent nasal, auricular or facial abnormalities. Her psychomotor development was normal. The single physiological distinction was that her right hand had six fingers and no thumb. Radiographs of the right forearm and hand showed ulnar duplication, absence of the radius and thumb, lower humerus articulating with two olecranon processes, and symmetric polydactyly (Figure 1). Her parents refused surgical treatment.

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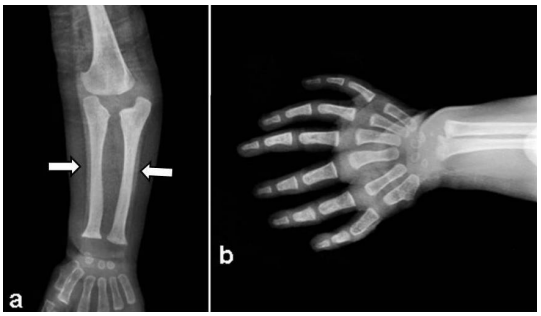

Figure 1
Radiographs of the right forearm (a) and hand (b) demonstrate duplicated ulna and six-finger symmetric polydactyly. There are two ulnae, with absent radius and thumb.

Discussion

The development of a duplicated ulnae indicates that altered signaling affected patterning early during limb bud outgrowth. Ulnar dimelia is not unique to humans. Gorriz et al. described the same phenomena in chickens [4], and Ueshima and Uehara described the condition in a calf [5]. It is usually sporadic, but Sandrow et al. described one family in which both the father and daughter were affected [6].The actual genetic basis for this condition is unknown, but a spontaneous mutation has been suggested. Ulnar dimelia affects both sexes indiscriminately. It is commonly unilateral, but a bilateral case was reported by Laurin et al. [7].

The classic "mirror deformity" involves complete reduplication of the ulnar ray. There are two ulnae, a duplication of the ulnar halves of the carpus, metacarpals and phalanges, and radius and thumb are absent. The upper limb anomaly is usually the only defect, but associated features including peculiar facies, congenital dislocation of the shoulder and fibular dimelia have been described [1-6].

A classification of the mirror hand-multiple hand spectrum in which patients had additional fingers has been proposed by Al-Qattan and Al-Thunayan et al. .In their classification, type 1 is associated with ulnar dimelia (two ulnae), type 2 is an intermediate with two ulnae and a radius, type 3 is another intermediate with one radius and ulna, type 4 is a syndromal mirror hand, and type 5 is multiple hands [8]. The case we are presenting is classified as type 1 in which the two ulnae are fully developed.

The embryology of mirror deformity was first described by Saunders and Gasseling, who produced mirror image digit duplications in chicks by grafting a small piece of the posterior border mesoderm into an anterior position [7]. This zone is known as the zone of polarizing activity (ZPA) and is regulated by the sonic hedgehog (Shh) gene, which is responsible for limb patterning on an anteroposterior axis. Ectopic expression of the zone of polarizing activity cells or the sonic hedgehog gene can therefore produce mirror image deformities. Secondary signaling molecules such as the Hox genes have also been implicated in the embryogenesis of mirror image deformities. Hox genes encode for positional information during embryogenesis. Hox b-8 is thought to be important in the specification of the zone of polarizing activity cell positioning, and ectopic expression of Hox b-8 has experimentally resulted in mirror image duplication [9-11].

In these patients, the stiffness of elbow and wrist are significant symptoms and remain difficult to remedy, whereas pollicization is now an established, successful mean by which to treat the hand deformity. The management of the ulnar dimelia deformity involves treatment of the elbow, wrist and hand [1-5].

In conclusion, congenital limb malformations may occur as an isolated trait, or in association with a malformation

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syndrome. Future genetic research may help to elucidate the causative pathogenesis of these types malformations.

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