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CASE REPORT

Combined Congenital Radial and Ulnar Longitudinal Deficiencies: A Case Report

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Introduction

The incidence of radial longitudinal deficiencies (RLD) is estimated 1 in 9,000 to 1 in 55,000 [1, 2]. The incidence of ulnar-sided defect or ulnar longitudinal deficiencies (ULD) is estimated 1 in 100,000 [1, 2]. Phocomelia is a very rare anomaly. Its prevalence was 5 in 4024000 in one series and a similar prevalence in the other series [3, 4]. Goldfarb et al. and Tytherleigh-Strong and Hooper suggested that phocomelia should be considered as proximal extensions of radial longitudinal dysplasia or/and ulnar longitudinal dysplasia [3, 4]. Combination of RLD and ULD in contra-lateral sides in the same patient is an exceedingly rare occurrence. [1] This case report describes a patient with right RLD and left ULD. The left ULD was extended proximally to become a phocomelic left upper limb.

Case Report

A 6-year-old boy was referred by welfare system for evaluation of his bilateral upper limbs anomalies. His parents were first cousins. His lower limbs appeared normal and he had no mental abnormality. His karyotype was normal. His mother's maternal history was not clear and she had no prenatal care. Evaluation of the heart, spine, kidneys and hematopoeitic system were normal.

The patient had a 4-fingered right hand which was radially deviated. The thumb was absent (Fig. 1). The right index finger was hypoplastic. The child performed grasp with his middle and ring fingers. Radiographs of the right upper limb showed that there were 4 triphalangeal fingers, lack of osseous structures of the thumb and absence of the radius (Fig. 2). Radius deficiency classified Bayne type IV [5] and thumb hypoplasia classified Blauth type V [6]. The child had a centralization procedure at 2 years old but the parents declined for pollicization.

The left upper limb was apparently a phocomelic limb. The forearm and hand were attached to the trunk (Fig. 1). There was no elbow joint. The left hand had 2 fingers. The left upper limb had no function. Radiographic evaluation demonstrated that the left upper limb osseous structure consisted of a small segment of humerus, a hypoplastic radius and abnormal carpal bones. The left hand had 2 triphalangeal fingers. The thumb was absent (Figs. 3 and 4). Left shoulder was under developed and unstable (Fig. 5). The forearm bone was not attached to the rudimentary humerus. There was no radiohumeral synostosis. Ulnar deficiency classified forearm/elbow Bayne type IV [7], absence of the ring and little fingers classified hand type D of the Ogino and Kato classification [8] and absent of the thumb classified type D of the Cole and Manske classification of ulnar deficiency according to the thumb and first web space [9]. The phocomelic left upper limb could not be classified by Frantz and O'Rahilly classification of phocomelia [3, 4]. However, it may be classified as a Proximal Ulnar Longitudinal Dysplasia of Goldfarb et al. classification of phocomelia [3], and described as type A (an abnormal humerus with an abnormal single forearm bone) Tytherleigh-Strong and Hooper classification of phocomelia [4].

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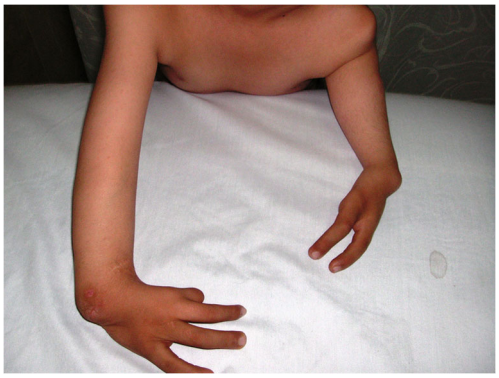

Fig. 1 Clinical photography of the child with right radial longitudinal deficiencies and a phocomelic left upper limb

Discussion

Limbs buds appear on day 26 of gestation. Development of limb bud occurs along the proximal-distal, antro-posterior and dorso-ventral axes. The apical ectodermal ridge (AER) is responsible for proximodistal development of the embryonic limb bud and secretes a group of fibroblast growth factors (FGFs) that regulate the normal limb outgrowth. The zone of polarizing activity (ZPA), located on the posterior margin of the limb bud, controls the radioulnar patterning through the mediation of sonic hedgehog (Shh) molecule. The complex interrelations between the ZPA and the AER are a part of a feedback loop. Shh signals from the ZPA regulate the activity of the AER. The AER gives positive feedback to the ZPA by FGF signals. ULD is thought to occur from disruption of ZPA formation or Shh dysfunction, while RLD is associated with disruption of AER-related cell proliferation [1].

The development of congenital abnormality is dose and time dependant. Ogino and Kato produced RLD and ULD in rats by administration of Myleran (Busulfan). The time of Myleran administration was critical to induce the type of dysplasia. Administration of Busulfan at 9 to 10 days of gestation produced ULD. Administration of Busulfan at 10 to 11 days of gestation produced RDL. The authors observed that the critical period to develop ULD was earlier than that required to produce RLD. The authors did not observe concurrent RLD and ULD [1, 8].

In the current case the patient's left upper limb had ULD with absence of the thumb and index finger. Radial side

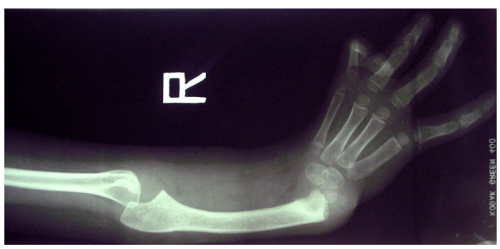

Fig. 2 Right upper limb radiograph demonstrates absence of the radius (Bayne type IV) [5] and absence of the thumb (Blauth type V) [6]

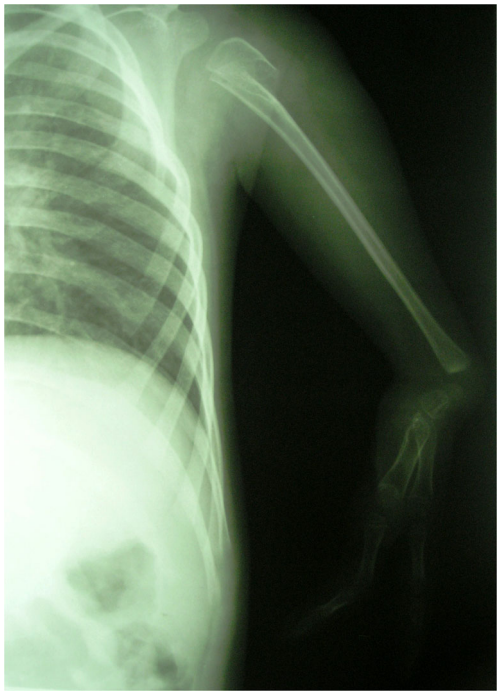

Fig. 3 Left upper limb radiograph demonstrates ulnar deficiency (forearm/elbow Bayne type IV) [7], absence of the ring and little fingers (hand type D Ogino and Kato) [8] and absence of the thumb (type D Cole and Manske classification) [9]. The phocomelic left upper limb could not be classified by the Frantz and O'Rahilly classification of phocomelia. However, it may be classified as a Proximal Ulnar Longitudinal Dysplasia of Goldfarb et al. classification of phocomelia [3], and describe as type A (an abnormal humerus with an abnormal single forearm bone) Tytherleigh-Strong and Hooper classification of phocomelia [4]

deficiencies have been reported in 68 to 100 % of ULD [9–11]. It has been speculated that an insult in the ZPA disrupts the normal development and differentiation patterns and produces deficiencies on both radial and ulnar sides of the same hand [9]. Radial side deficiencies have been elaborated in the classification of ulnar deficiency according to the thumb and first web developed by Cole and Manske [9].

Usually patients with bilateral congenital anomalies have the same anomaly on both sides; however, the severities may be varied [3, 4]. Combination of RLD and ULD on contra-

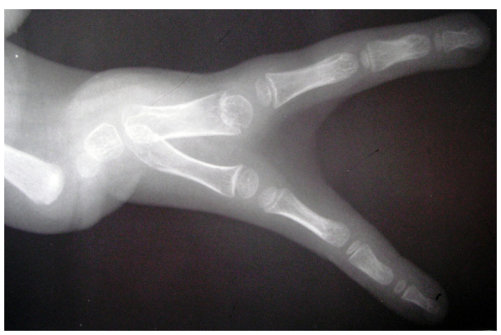

Fig. 4 Left hand radiograph demonstrates absence of the ring and little fingers (hand type D Ogino and Kato) [8] and absence of the thumb (type D Cole and Manske classification) [9]

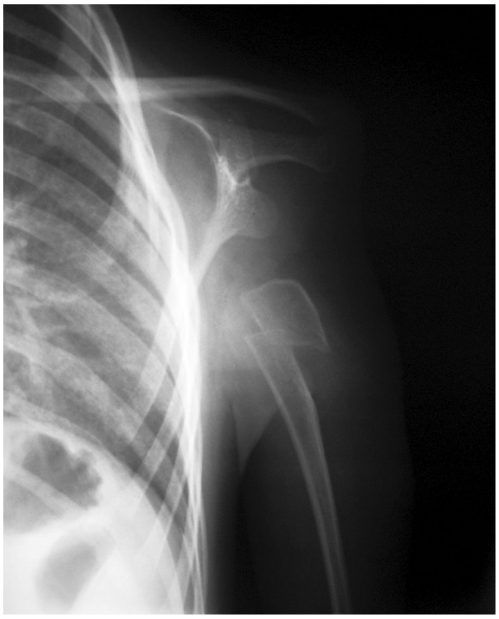

Fig. 5 Left shoulder is under developed and unstable. The forearm bone is not attached to the rudimentary humerus

lateral sides in the same patient is an exceedingly rare occurrence. I was able to find only a report of two cases describing this occurrence [1]. Oberg et al. have reported this combination in a fetus and a newborn. The fetus's mother maternal history was unclear; however, the newborn's mother had a distinct history of 2 bleeding episodes during pregnancy, the timing of which was probably correlated with the occurred RLD and ULD. Further follow up of the newborn was not provided [1].

In the current case as well as the two reported cases by Oberg et al. RLD was on the right side while ULD was on the left side. Collective data suggest that ULD is more common on the left side while RLD is more common on the right side [1]. Oberg et al. suggested that the sidedness preference might be due to the role of Shh role in establishing left-sided asymmetry during gastrulation. This may demonstrate a regulatory complexity for Shh on the left side that is more easily disrupted leading to increases susceptibility for ULD in the left limb [1].

In the current case, I suspect that the combination of RLD and ULD on contra-lateral sides is due to two separate teratologic insults at two different times during embryonic development. The first insult affected the left limb bud that led to ULD with proximal extension and the second insult affected the right upper limb that led to RLD. [1, 8, 12] However, further investigation is needed to substantiate these hypotheses for development of this rare combination of the anomalies. This case is presented because of its rare occurrence and each reported case will hopefully bring more insight to the congenital anomalies.

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