Ulnar aplasia, dysplastic radius and preaxial oligodactyly: Rare longitudinal limb defect in a sporadic male child

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Ulnar hypoplasia is a rare longitudinal limb deficiency in which the ulna shows various degrees of deficiency. The condition is normally associated with radial defects, and in severe cases there is a reduction of postaxial/unlar digits. Ulnar deficiency is an integral part of several syndromic malformations like Weyer’s oligodactyly syndrome, limb/pelvis hypoplasia/aplasia syndrome, and ulnar-mammary syndrome. Here, we report an isolated unilateral ulnar deficiency in a boy who was a product of a consanguineous marriage. The subject demonstrated mesomelic shortening of the left arm with reduced zeugopod and autopod, and preaxial absence of two fingers. Additional findings in the affected limb were severe flexion contracture at the elbow joint, reduced and narrow palm, hypoplastic digits, and clinodactyly. Roentgenographic study revealed rudimentary ulna, dysplastic and posteriorly dislocated radius, crowding of carpals, and complete absence of digit rays of the thumb and index finger. Despite this anomaly, the subject could manage his daily life activities well. We present detailed clinical features and differential diagnosis of this rare limb malformation.

Key words: Finger reduction, longitudinal defect, limb deficiency, oligodactyly, Pakistani subject, radial dysplasia, ulnar aplasia

CASE REPORT

The subject, a 12-year-old school-going boy, originates from a rural area of Southern Punjab, Pakistan. His parents were first cousins (inbreeding coefficient, \(F = 0.0625\)), and he had four normal siblings (3 brothers, 1 sister). The maternal and paternal ages were 28 and 33 years, respectively, at the time of his birth. The pregnancy had been uneventful and the birth was at home, in the presence of a traditional birth attendant. The study was approved by the institutional review committee and all the information was obtained according to the Helsinki II declaration. The initial ascertainment and detailed clinical examinations were carried out in several visits during 2009-2010.

The subject had normal developmental landmarks, dentition, and intelligence quotient (IQ). He was observed to have an isolated limb anomaly. Upon physical examination, he had a standing height of 146 cm, sitting height 71 cm, arm span 130 cm, head circumference 53 cm, neck circumference 29 cm, and chest 68 cm. There was no family history of any limb or other anomaly.

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Clinical examination showed a short left arm [Figure 1A]. The upper arm was thin and weak, and the middle and forearm were reduced in size. The elbow demonstrated limited flexion and extension [Figure 1A,B]. The middle arm was flat and depicted restricted supination. The palm was narrow with a few flexion creases [Figure 1C]. The ‘hand’ harbored only three functional fingers identified as third, fourth, and fifth, each comprising two flexion creases at the interphalangeal joints. The thumb and index finger were completely omitted. The posterior finger, that is, the fifth digit, was weak and showed volar inclination [Figure 1A,C].

In the roentgenograms, the ulna was grossly degenerate and was only represented by a club-shaped decalcified bony island of ~2.5 cm at the distal zeugopod [Figure 1D, E; Table 1]. The radius appeared short, hypoplastic, and dysplastic. In the absence of the ulna, the radius demonstrated a compensatory curvature and maintained a varus concavity of ~20° [Figure 1D]. Its proximal head was ill grown, misaligned, and posteriorly dislocated, and was losing its normal axis along the humeral trochanter. The distal head of the radius was broad, weak, and decalcified, depicting signs of immaturity, and was capped with dysplastic epiphysis [Figure 1D]. In the upper arm, the humerus was short, thin, and decalcified [Figure 1B; Table 1]. Its distal head was dysplastic with ill-developed epiphysis and hence, was unable to support the zeugopod.

Figure 1: Phenotype in subject; a-e: Photographs and radiograph of upper limbs
In the autopod, four carpals were visible which were crowded and malformed [Figure 1E]. Three metacarpals were present which though thin, maintained normal size [Table 1]. At the anterior axis, the ‘third’ finger depicted volar inclination, whereas at the posterior axis, the ‘fifth’ digit showed clinodactyly [Figure 1E]. There was minor cutaneous syndactyly between the fourth and fifth fingers which was not witnessed at the osseous level [Figure 1C] (see Table 1 for detailed roentgenographic measurements).

In the right arm, roentgenograms were unremarkable except for crowding of the carpals. There was no involvement of any other organ system; there were no symptoms of oral, faciocranial, skeletal, or vital internal organs. His parents could not recall any drug exposure during the pregnancy. The subject has fully adapted this limb deficiency and can manage his daily life activities well.

**DISCUSSION**

Ulnar longitudinal deficiencies are very rare and their prevalence has been estimated to be 1-2 in 100,000 live births. Several different classification systems have been proposed for ulnar ray deficiency (reviewed by Al-Qattan, 2010). O’Rahilly suggested a classification of congenital limb anomalies that was based on anatomical differences. Ogden et al. provided a classification based upon the severity of the ulnar deformity. Bayne considered the degree of ulnar deficiency and also the radiohumeral synostosis. Ogino and Kato provided a system which also integrated into the classification the deficiency of fingers at the ulnar side. Cole and Manske added thumb/first web deformity to the ulnar ray deficiency. Hence, according to the O’Rahilly scheme, the present anomaly can be classified as ulnar-preaxial carpal-phalangeal meromelia. According to the Bayne system, the presentation in our subject is close to type II (i.e., partial aplasia of the ulna). On the other hand, our case is difficult to characterize according to the Ogino-Kato scheme because here the radial digits are omitted and the remaining fingers maintain posterior/ulnar identities. According to the Cole-Manske system, which also considers the absent thumb, the present case is closer to type D. Proximally, when the ulna is absent, the radius can be unstable in its articulation with the humerus and shows proximal dislocation. This situation was appreciated by Kummel who provided a classification of elbow deformity. Accordingly, our case can be classified as type III (i.e., radial head dislocation).

The clinical presentation in our subject is best classified as ulnar aplasia, dysplastic radius, and preaxial oligodactyly. There was no evidence of radioulnar synostosis or fusion at the angle joint, that is, humeroradial juxtaposition. However, severe flexion contracture of the elbow and radial head dislocation were witnessed. The radiocarpal joint was stable. We also considered differential diagnosis and reported cases with the Schinzel syndrome, Marles-Chudley syndrome, Weyer’s ulnar ray/oligodactyly syndrome [Online Mendelian Inheritance in Man (OMIM): 602418], limb/pelvis-hypoplasia/aplasia syndrome (OMIM: 276820), ulnar-mammary syndrome (OMIM: 181450), Fuhrmann syndrome (OMIM: 228930), and Cornelia de Lange syndrome (OMIM: 122470). However, all these syndromic anomalies can be safely excluded as they show associations of orofacial, cardiac, and/or ectodermal organs.

The studies on hereditary and congenital limb deficiencies are relatively scarce in Pakistan. Only a few reports depicting digit deficiencies have been published. There is no previous study on longitudinal limb defects.

Congenital limb and/or finger loss is a potentially devastating event which imparts profound physical, psychological, and vocational consequences on the subject as well as his/her family. Despite the potential adverse...
impact of partial loss or deficiency on daily function and the quality of life, management is not easy and no immediate help is available. Prosthetic and cosmetic measures not only require long-term follow-up and skilled surgical procedures but are also known to offer limited rescue. The etiology of such defects is not fully understood mainly due to the rarity of reported subjects and familial cases, poor understanding of inheritance patterns, and lack of molecular and genetic data. Further studies are warranted on the pathophysiology of longitudinal limb defects to propose better therapeutic and surgical options for the subjects.

Author contributions
MA ascertained the case and helped in data collection. SM analyzed the data and drafted the manuscript. Both authors read and approved the content of the manuscript.

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