



Ulnar Hemimelia with Humero-Radial Synostosis and Oligodactyly: A Rarity and Review of Literature

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Abstract

Ulnar hemimelia is a very rare condition. In most of the cases, it goes unreported due to death of fetus in-utero owing to other anomalies during organogenesis. The association of ulnar hemimelia with humero-radial synostosis and oligodactyly makes our case very rare and unusual. It presents as shortening of upper limb, rigid elbow, ulnar clubbing and coalition or absence of metacarpals. A rare case of ulnar hemimelia with humeroradial synostosis and absence of one metacarpal on ulnar side is presented in an 8 months old female child. Detailed review of literature Management of this condition is quite individualized. The management of this condition involves early physiotherapy followed by multiple soft tissue release if required. Parental counseling is of utmost importance while dealing with such conditions.

Introduction

Ulnar hemimelia (ulnar club hand, ulnar ray deficiency) was described by Swanson et al. [1] in 1984 as partial or complete failure of the formation of ulna. It presents very rarely and reported incidence is 1 in 150,000 [2]. These types of anomalies develop earlier in embryonic life as compared to other skeletal anomalies and this may be the reason for its rarity. Embryologically, the most critical period for the development of these limb anomalies is from 24 to 36 days of embryonic life [3,4]. Ulnar hemimelia is more common in men, with a male to female ratio of 3:2 and it is unilateral in about 70% cases and mostly right sided [2,3]. During embryonic period some environmental factors such as history of smoking, cocaine use and use of teratogenic drugs by mother may cause this anomaly [5]. The ulnar hemimelias are mostly congenital and reported as partial absence of ulna. But sometimes they may be associated with other skeletal anomalies such as humero-radial synostosis, radial head dislocation, carpal or metacarpal coalition, and digital abnormalities [6]. Previously few such cases have been reported in the literature [1,7-9]. Informed consent was taken from patient's mother regarding publication of case related data and study was approved by the Institute ethics committee. We present a rare case of ulnar hemimelia with humeroradial synostosis and absence of one metacarpal on ulnar side in an 8 months old female child. Detailed review of literature of ulnar hemimelia with humero-radial synostosis and oligodactyly is also presented and discussed.

Case Presentation

An eight months old female child presented to our clinic with complaints of short left upper limb and restricted activities. The antenatal period and the delivery were uneventful. The pregnancy was not followed up by any health care worker and the delivery took place at home. There was no history of drug intake or smoking by mother during her pregnancy. On examination, the left upper limb was found to be shorter than the contra-lateral limb (Figure 1a and 1b). The shoulder, wrist and finger movements were found to be within normal limits. However, there was no movement at elbow. Other findings included bowing of forearm, elbow fixed in 90 degrees of flexion and quadridactyly with absence of the little finger. This limb was mal-rotated with the elbow being anterior and the cubital fossa posteriorly. Laboratory investigations were within normal limits. Radiographs showed left upper limb micromelia, humero-radial synostosis and only a single bone was present in the forearm (Figures 2a-2c). The carpal bones were incompletely ossified but there were only four metacarpals and four phalanges. The hand was deviated ulnarward. She had no detectable overlying skin abnormality in this limb. Whole abdominal ultrasonography revealed no renal or other anomalies. The echocardiography of this patient showed no cardiac anomalies.

Discussion

Ulnar hemimelia or ulnar ray deficiency is one of the longitudinal deficiencies of the upper

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Figure 1a: Clinical photograph of the child showing Left upper limb congenitally shortened, rotated with elbow facing posteriorly and ankylosed in 90° flexion.



Figure 1b: Clinical photograph of the child showing Left upper limb congenitally shortened, rotated with elbow facing posteriorly and ankylosed and oligodactyly.



Figure 2a: Photograph showing X-rays of left upper limb Anteroposterior view showing Radio-humeral synostosis with ulnar hemimelia and oligodactyly.

limb. Congenital ulnar hemimelia results due to longitudinal failure in formation of part or whole of a long bone. According to Ogino and Kato [4], the critical period of ulnar development is earlier than that of other bones. Any abnormality during this period leads to mortality of fetus. This explains why ulnar deficiency is rarely reported. Ulnar deficiency is mostly non-syndromic but it may also be present with Poland syndrome, Goltz-Gorlin syndrome, Cornelia De Lange syndrome, or femur fibula ulna syndrome [10]. In our patient presentation of unilateral ulnar hemimelia associated with humero-radial synostosis, malrotated elbow and four fingered hand makes it more complex and unusual variant. Moreover it presented in the female in the present study which is unusual to the more prevalence



Figure 2b: X-rays of left upper limb Lateral view showing Radio-humeral synostosis with elbow fixed in 90° flexion, ulnar hemimelia and oligodactyly.



Figure 2c: X-rays of left upper limb Lateral view showing Radio-humeral synostosis with radial bowing, ulnar hemimelia and oligodactyly.

in males [2]. Bayne [11] classified ulnar hemimelia into 4 types based on radiology. Type 1 involves hypoplasia of the ulna where both distal and proximal ulnar epiphyses are present. Type 2 is the partial aplasia of the ulna. Type 3 is the complete absence of the ulna that accompanies severe carpal and digital deficiencies. Type 4 includes humero-radial synostosis. Humero-radial synostoses are rare and current classification divides the synostosis into Class I (fixed in extension with ulnar ray hypoplasia and sporadic) and Class II (fixed in flexion without hypoplasia and familial) [12]. Ulnar hemimelia may be associated with complex carpal, metacarpal, and digital abnormalities. There may be an associated aplasia, hypoplasia, and fusion of carpal or metacarpal bones. Among carpals, the triquetrum and the capitate are commonly absent in these patients. The most common hand anomaly associated with this anomaly is three-fingered hand (tridactyly), followed by mono-digital hand [6,8]. In our patient, ulnar hemimelia of type 4 and humero-radial synostosis type does not fit into any of type (Table 1). Swanson et al. [1] reported that 47/88 limbs (53.4%) associated with humero-radial synostosis, 21/88 limbs (23.9%) with total absence of ulna, ulnar deviation of wrist in 22/88 limbs (25%) and about 90% patients with 1 to 4 digits in hand. Study by Elhassan et al. [7] reported that in all patients of type IV ulnar hemimelia with humero-radial synostosis, elbow was fixed in 10 to 90 degree of flexion. Pfeiffer and Braun-Quentin [13] studied genetic basis of humero-radial synostosis and classified it into 3 entities as: (1) autosomal dominant humero-radial synostosis with multiple joint synostosis; (2) autosomal recessive humero-radial synostosis with dysgenesis of the ulna and possibly the fibula and femur but without digital anomalies; and (3) non-germinal humero-radial synostosis as part of ulna longitudinal dysplasia with digital anomalies. Humero-radial synostosis may also be associated with a rare Antley-Bixler syndrome, in which elbow is fixed in about 90 degree of flexion with other anomalies that includes camptodactyly,

Table 1: Clinical manifestations in Bayne's type IV ulnar hemimelia.

Site	Clinical manifestations	Our case findings
Shoulder girdle	Normal & stable or glenohumeral dysplasia & unstable	Normal; stable
Elbow	Humero-radial synostosis; fixed in 10° to 90° flexion or fixed in extension; humerus internal rotation & forearm pronation	Humero-radial synostosis; fixed in 90° flexion; forearm completely pronated
Radius	Radial bowing	Severe radial bowing
Ulna	Complete absence; analge present	Complete absence
Wrist	Ulnar deviation; carpal aplasia, hypoplasia or fusion	Ulnar deviation present
Hand	1-4 digits, absence of all digits	Oligodactyly; four fingered hand
Other skeletal manifestations	Proximal femur focal deficiency, fibular ray deficiency	None

arachnodactyly, joint contractures, and craniosynostosis, dysplastic ears, midface hypoplasia, and choanal atresia [14]. Our patient did not have Antley-Bixler syndrome because of isolated unilateral upper limb involvement. Management of these patients is quite challenging owing to the complex nature of anomalies and depends upon individual condition of patient. It begins with early passive physiotherapy of limb to avoid disuse atrophy at later stage. Most of these unilateral ulnar hemimelias are managed without surgical intervention [8]. Surgical procedures, if required, are based on age and deformities present in the patients. Multiple soft tissue release and humeral de-rotational osteotomy is an option that may help to reduce disability of the patient. In a study by Lovett [15] on treatment of longitudinal ulnar deficiency, out of total 61 patients, humeral rotational osteotomy was performed in 2 patients and elbow Z-plasty in 3 patients. Although, cosmetically and functionally humerus rotational osteotomies are gives better results. In majority of patients with single limb involvement no significant treatment was recommended. In our case we started the treatment with continuous passive physiotherapy. We also counseled the parents regarding progress of the condition. We hope to follow the patient until adulthood and may need to have some surgical intervention to free the elbow joint.

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