Ulnar Hemimelia with Oilgodactyly: Report of Two Cases

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Abstract

We present two sporadic cases of complete ulnar hemimelia, a rare congenital defect. In one case, ulnar hemimelia was associated with tridactyly and elbow malrotation with radiohumeral synostosis; in the second case, ulnar hemimelia was associated with mono-metacarpal bidactyly and anterior cubital webbing of the elbow with fixed flexion. To the best of our knowledge, there is dearth of information on these combinations of anomalies in the literature and their management remains a challenge.

Introduction

Isidore Geoffroy Saint-Hilaire coined the term “hemimelia” in the early 19th century, while in 1951, O'Rahilly suggested the term "paraxial hemimelia" for the longitudinal variety, because either the preaxial or postaxial side of the limb is involved [1]. The deficiency can be transverse, longitudinal and sometimes combination of both [1-5]. Transverse deficiencies results in congenital limb amputation and is more common in males with a gender ratio of 3.5:1 [3,4]. The left limb is twice involved than right limb and the upper limbs are affected more than lower limbs [1,4,5].

In the upper limb longitudinal deficiency, a bone of the forearm fails to develop in part or in whole. It may be preaxial (radial ray deficiency or radial club hand) or postaxial (ulnar ray deficiency or ulnar club hand). Radial club hands have an incidence that varies between 1:55,000 to 100,000 live births, while the less common ulnar hemimelia occurs in about 1 in 150,000 [1, 5].
Hemimelias are more often than not congenital. However, there have been rare reports of acquired hemimelia [6, 7]. Embryologically, most of the morphological differentiation of the limbs occur during the embryonic period, which end by the 8th weeks. The most critical period for the development of limb anomalies is from 24-36 days of embryonic life [1,6,8]. However, ossification and growth proceeds throughout the fetal period to puberty. Hence, congenital anomalies of the extremities are mainly genetic in origin, but some environmental factors such as maternal cigarette smoking and some drugs used during pregnancy have been implicated [1,5,7].

Clinically, because of the multiplicity of forearm and hand deformities or contours, it may be very difficult to recognize precisely the deficiency without radiological studies [1]. In the prenatal to early neonatal life, sonography is a valuable imaging modality for diagnosis and determination of organ anlage. Radiography is diagnostic and is of value at excluding VACTERL (vertebral, anal, tracheal, cardiac, esophageal, renal, and limb abnormalities) association where present. Magnetic resonance imaging has wide range application at demonstrating organ anlage and vascular malformations but for cost may be reserved for cases that are inconclusive with other studies.

This reports details the findings in two very rare cases of upper limb hemimelias combining radiohumeral synostosis, elbow malrotation and tridactyl in one, and fixed flexion with cubital webbing of the elbow, mono-metacarpal and two-finger hand on ulnar ray deficiency in the other.

Case Reports

**Case 1:** A 5-month-old boy whose pregnancy, labor and delivery were uneventful was presented to the outpatient department of our hospital with complaint of small left upper limb with poor activities. He is the second child (both alive) of a 21-year-old woman. Pregnancy was booked at 26 weeks. Ultrasound scan at book was reported as normal. There was no history of drug ingestion other than the routine antenatal drugs, which are folic acid, chloroquine and ferrous sulphate. Apgar score at birth was estimated at 7, 10, and 10.

On examination, the left upper limb was smaller and shorter than the contra-lateral limb, showed poverty of movement and but fair hand gripping, rigid elbow, radial clubbed hand and tridactyly with absence of the ring and little finger. This limb was mal-rotated with the elbow being anterior and the cubit posteriorly (Fig. 1). There was no other phenotypical abnormality.

Laboratory work-up, including full blood count, was normal. Abdominal ultrasonography excluded renal anomaly. The clinical examination, echocardiography and electrocardiography of this patient showed no cardiac abnormality. Radiographs (Fig. 2) showed left upper limb micromelia.

The left shoulder and the humerus, although smaller than the contralateral side, were essentially normal. However, only a single bone was present in the forearm. Its proximal end was locked into the humeral metaphysis at the midline like a ‘peg in a hole’. This bone could not be identified with certainty as radius or ulna. There was bowing of its shaft convex anteriorly. The carpal bones are incompletely ossified but there were only three metacarpals and three phalanges. The hand was radially deviated (radial clubbing). He had no detectable overlying skin abnormality in this limb.
Figure 1. Case 1. Baby boy with hemimelia. Photograph at age 5 months shows left upper limb hemimelia, shortened left upper limb, malrotated elbow, and three-fingered left hand.

Figure 2. Case 1. Baby boy with hemimelia. (A-B) Radiographs show left upper limb ulnar hemimelia. There is a normal right upper limb but complete absence of the left ulna. There is radiohumeral synostosis and a three-fingered hand.

Case 2: A 3-month-old male infant was delivered to a 28-year-old para 2 mother, both alive. Mother had antenatal care at a maternity home with limited facilities. Thus, she had no antenatal ultrasound scan. Mother was not on any drug with known teratogenic effect. She neither smokes nor drinks alcohol. Laboratory and radiological work up in this case were essentially normal. However, fixed elbow flexion, anterior cubital webbing, and one metacarpal with two-fingered hand in this case characterized the left limb deformity (Figs. 3 and 4).
Figure 3. Case 2. Baby boy with hemimelia. Photograph of at age 3 months shows left upper limb hemimelia, elbow webbing in fixed hyperflexion, and a two-fingered left hand.

Figure 4. Case 2. Baby boy with hemimelia. (A-B) Radiographs show left upper limb ulnar hemimelia. Note the normal right upper limb and complete absence of the left ulna, with fixed flexion deformity of the elbow, mono metacarpal and a two-fingered hand with ulnar ray deficiency.

Discussion

Longitudinal failure of formation of parts or whole of a long bone in the upper limb resulting into congenital radial hemimelia and ulnar hemimelia when the radial and ulna bones respectively are completely or partially absent is rare [3,4]. The critical period of ulnar deficiency, according to Ogino and Kato [8] is earlier than that of other anomalies and it corresponds to the period of a high mortality rate of fetuses. This may explain why ulnar deficiency does not appear as often as other anomalies. In our patients, the ulnar clubbing of the hands and the presence of thumb in them negated radial ray
deficiency. The radio-humeral synostosis from failure of cavitations and the elbow mal-rotation in the first case makes the anomalies more complex (Fig. 2a). In few instances associated with radio-humeral synostosis, there was associated anterior cubital webbing as in Case 2. Thus, Case 1 having mal-positioned cubit is the more unusual variant of ulna hemimelia.

Ulnar deficiencies are often associated with complex involvement of carpals, metacarpals and digits [1, 8, 9]. The triquetrum and capitate often are absent and there is an increasing frequency of metacarpal failure from the radial to the ulnar side of the hand and high frequency of digital absence with three-fingered hand predominating followed closely by the mono-digital hand [1, 9]. Case 1 with tridactyly conforms to this description, but the mal-rotated ‘locked elbow’ (radio-humeral synostosis) into anterior position, appearing as ‘peg in hole’ is rarely reported. The degree of carpal deficiencies in our cases is still radiographically uncertain because of the incomplete ossification expected at their young ages. The case of an una hemimelia with radio-humeral synostosis in Frantz and O’Rahilly’s series [1] who also had three-finger-hand in addition to elbow mal-rotation is similar to our Case 1. However, we found no case of ulnar hemimelia having mono-metacarpal with two-finger hand as in our Case 2. In the work of Elhassan et al [9], 12% of cases of ulnar hemimelia demonstrated radiohumeral synostosis and 65% of patients with radio-humeral synostosis have digital anomalies.

The unilateralism of hemimelia in our patients has been variably reported by many authors to occur in as much as 80% of hemimelia and confers better prognosis in term of functionality [10]. The ‘locking’ of the proximal portion of the only forearm bone in Case 1 into the distal humeral metaphysis midway between the epicondyles is unusual for either ulna or radio-humeral articulations.

Ulnar hemimelia is rarely associated with syndromes unlike the radial clubbed hand that has been associated with many congenital syndromes including Holt-Oram syndrome (cardiac septal defects), thrombocytopenia with absent radius, Fanconi anaemia (aplastic anaemia), and VACTERL syndrome [1, 10, 11]. We are uncertain whether our cases are syndrome complex because of lack of information on these types of anomalies in the electronic literatures searched to the best of our knowledge.

According to Shafi and Hui [10], where hemimelia is associated with a syndrome, the prognosis depends on it. There was no anomaly identified in the other limbs and elsewhere in our cases on clinical examinations (bedside and laboratory) and radiological work up (using radiography and sonography). Again, the absence of contributory family and drug histories in addition to the non-specific linkage to any environmental factor in these patients led us to belief these combinations of abnormalities as sporadic occurrence. Radiographs can be useful in assessing the degree of bony aplasia, while laboratory analysis that need to includes tests for anaemia and thrombocytopenia are invaluable in the patient diagnostic work up. Laboratory work excluded thrombocytopenia with absent radius syndrome and other forms of anaemias in our patients. In addition, the absence of evidence of cardiac abnormality on both echocardiography and electrocardiography in our patients make Holt-Oram a remote possibility.

The management of these patients for optimal function is a dilemma because of the oligodactyly, elbow deformities and their young ages. Generally, the management of upper limb hemimelia is challenging and is largely individualized [1, 11, 12]. The timing and procedure to be performed may be dictated by the age of the patient, bilaterality, and the scope of the handicap. Most of the affected children are managed without surgical intervention especially where affectation is unilateral. The goal of intervention is to improve function with or without the use of prosthesis [1]. The several non-surgical management available for the varying degrees of hemimelia includes opponens post, the use of above elbow prosthesis and below elbow prosthesis. These are usually applicable when the patient
is grown up, from adolescence age. The option of prosthesis is not been consider for now in our patients due to their young ages in addition to financial limitation.

Surgery may be highly indicated in bilateral affectation and significant reduction in the range of movement [1,11]. The number of digits present, the degree of limb atrophy and the presence of syndactyly could also be a strong factor for surgery [1,11]. Numerous surgical options are available with varying limitations and advantages [1]. Some surgical interventions have been found to be rewarding. The use of Z-plasty in the cubital fossa to decrease the cubital web and improve the range of elbow flexion and extension allows a better fit to the forearm socket. However, it has failed significantly to increase the range of motion and therefore not usually recommended. The use of elbow disarticulation with an outside locking of elbow is more acceptable because it offers two different positions of the elbow joint. Humeral derotation osteotomy is an option that could help improve the range of movement across the joint surface.

We however, have some peculiar limitations in this cases originating from a resource-limited environment where good-fitting prostheses are not readily available and affordable, and both parents are junior civil servant who may not be able to afford good prosthesis. However, there is some luck in our patients, both having an anomaly free right upper limb. These babies are being train to make maximum use of the right limb, which we hope will be without serious handicap to their livelihoods. Training is ongoing in both patients on making maximum use of the normal right limbs and to probably improve functioning of the affected limbs and reducing the degree of atrophy from non-use.

We hope to follow them up until adulthood; they may however need to have some surgical intervention along the way to free the elbow joint and a well fitting prosthesis for the forearm and hand. These options are being considered because we are aware disused atrophy may set in with time and may make their abnormalities rather more obvious. Continuous physiotherapy, which we have started early in these cases, may assist and help to reduce the degree of atrophy from non-use.

References

1. Frantz CH, O'Rahilly R. Ulnar Hemimelia. Artificial Limbs. 1971; 15: 25-35. [PubMed]

2. Swanson AB. A classification for congenital limb malformations. J Hand Surg Am 1976; 1:8-22. [PubMed]

3. Jain SK, Lakhtakia PK. Profile of congenital transverse deficiencies among cases of congenital orthopaedic anomalies. Journal of Orthopaedic Surgery 2002, 10: 45–52. [PubMed]

4. Jain SK. A study of 200 cases of congenital limb deficiencies. Prosthet Orthop Int 1994, 18: 174–9. [PubMed]

5. Rogala ET, Wynne-Davies R, Littejohn A. Congenital limb anomalies: frequency and aetiological factors. Data from the Edinburgh Register of the Newborn (1964-68). J Med Genet 1974;11: 221. [PubMed]

6. Kanojia RK, Sharma N, Katariya H. Acquired radial club hand with humeroulnar dislocation: a rare sequel to infantile compartment syndrome following venous cannulation: a case report. Journal of Orthopaedic Surgery 2007;15:109-12. [PubMed]

7. Dreyfuss U. Acquired club hand--a case report. Hand 1977;9:268–71. [PubMed]
8. Ogino T, Kato H. Clinical and experimental studies on ulnar ray deficiency. Handchir Mikrochir Plast Chir. 1988;20:330-7. [PubMed]

9. Elhassan BT, Biafora S, Light T. Clinical manifestations of type IV ulna longitudinal dysplasia. J Hand Surg [Am]. 2008;33:617. [PubMed]

10. Shafi M, Hui JHP. Common paediatric orthopaedic problems in the upper limb. Singapore Med J 2006; 47: 654-659. [PubMed]

11. Johnson J, Omer GE Jr. Congenital ulnar deficiency. Natural history and therapeutic implications. Hand Clin. 1985;1:499-510. [PubMed]

12. Cole RJ, Manske PR. Classification of ulnar deficiency according to the thumb and first web. J Hand Surg [Am]. 1997;22:479-88. [PubMed]

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