A Case of Ulnar Hemimelia -- Rare Anomaly

Dr. Sangeeta J Rajani *, Dr. Jitendra K Rajani, Dr. Paresh Golwala*****

*Associate Professor, Department Of Anatomy, Medical College Baroda, vadodara.**Professor & Head, Dept Of Oral Pathology, Faculty of Dental Sciences, Dharamsinh Desai University, Nadiad.,***Professor & Head, Dept Of Orthopaedics, SBKS Medical Institute & Research Center , Pipariya, vadodara, Gujarat, India

Abstracts: When one side of the distal half of limb [leg or forearm] is absent, such cases are termed as hemimelia. Ulnar hemimelia is a very rare congenital anomalies, occurs in about 1 in 150,000. It is characterized by partial or complete absence of ulna, radial bowing, fixed or mobile elbow with abnormal digits. Most of the cases are unilateral [67%], mainly involves right side [69%] and more common in males [2:1]. In our case, a working boy presented with deformed right upper limb with tridactyly and elbow was fixed in extension. Xray examination revealed partial absence of ulna. The most critical period for the development of limb anomalies is from 24-36 days of embryonic life. Hence early diagnosis in antenatal period can reduce the occurrences. Management of such cases is highly individualized and mainly involves the improvement of function. In this case boy was independent in his daily routine activities hence no treatment required.c
[Rajani S et al NJIRM 2012; 3(3) : 190-194]
Key words: absent ulna, elbow synostosis, limb anomaly.

Author for correspondence: Dr. Sangeeta Jitendra Rajani, Associate Professor, Department of Anatomy, Medical College Braoda, (Gujarat). E-mail address: sangeetarajani73@yahoo.co.in

Introduction: Isolated deficit of long bones form a well recognized group of congenital anomalies. When one side of the distal half of limb is absent, such instances were named (after the defective portion) as hemimelia, and such instances were named “Ulnar”, “Radial”, “Fibular” and “Tibial” hemimelia. The term ‘hemimelia’ was introduced in 1836-37 by Isidore Geoffroy Saint-Hilaire. In 1877, Verneuil proposed subdivision of "ectromelia"(i.e. partial absence of limb) into longitudinal and transverse varieties. O'Rahilly[1] suggested the term "paraxial hemimelia for the longitudinal variety, because either the preaxial (radial ray deficiency or radial club hand) or postaxial (ulnar ray deficiency or ulnar club hand) side of the limb is involved. Ulnar hemimelia is a postaxial longitudinal deficiency of the Upper Limb, where in the ulna is completely or partially absent. The error is due to longitudinal suppression and involves the forearm bones and the developmental suppression continues distally to include the radial or ulnar rays of the metacarpals and fingers. Sometimes defect doesn’t affect the digits but it involves one or more carpal bones and fingers intact.

Classical presentation of ulnar hemimelia shows a partial rather than complete absence of the ulna with radial bowing, radius is proximally subluxated or fused with humerus and hence elbow fixed either in flexion or in extension position with abnormal digits. Studies by A R Southwood [2] reveal the Kummel’s classification regarding congenital ulnar defects into three groups-

1. Ulna partly or completely absent, the Radius normal though curved and ulnar fingers absent.
2. Ulna partly or completely absent, the Radius is ankylosed with the Humerus at more or less obtuse angle. If a proximal part of ulna is present, it is fused with radius and humerus.
3. Ulna partly or completely absent, the proximal end of Radius dislocated forwards and upwards on the Humerus.

Arthur Steindler[3] mentioned two types of Ectrodactyly (absence of phalangeal ray), central and marginal. Central where central ray is absent and in marginal, absence of the thumb or little finger. Marginal ectrodactyly is found in combination with absence of the one of the forearm bones.

Aim of this study is understand the classical features of the condition and find out the frequency of appearance of various features and its developmental etiology.

Case Report: A 25 years old, working boy visited dental OPD for routine check up and we noticed that he is having deformed right upper limb. Right upper arm was shorter than the left, associated with tridactyly [only three normal sized fingers]. On request, he allowed us for examination.

On examination, muscular development of right upper limb was poorer but in other respect he was well developed. The short Right upper limb, rest at the side of thorax, with fixed elbow in extension, forearm showed medial rotation on the Humerus [Figure 1].

He had normal right shoulder functions and fair hand grip, he was able to abduct and forward-flex the shoulder which allow him to do daily routine activities. Wrist permitted flexion through a range of 30 degree and there was lack of pronation. The bony structures of forearm were covered by the thick mass of muscles and are hypertrophied.

Figure 1 25 year boy with short and deformed right upper limb. Figure 2: Right hand with tridactaly and rudimentary finger on ulnar side

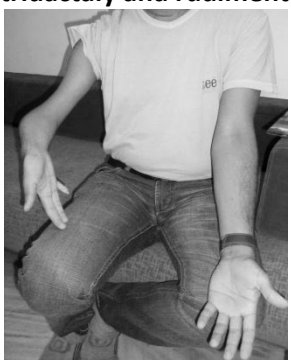

FIG 1

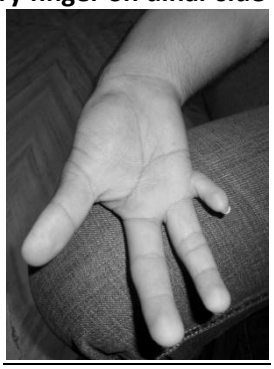

FIG 2

Hand was deviated to the ulnar side and making an angle of around 20 degree with the forearm. Hand consists of Thenar and Hypothenar eminence, and three normal sized fingers [Thumb, Index and Middle] and one rudimentary finger on ulnar side [Figure 2]. The condition provided an example of Kummel's second group. Minoo patel[4] revealed the studies of Elliott et al and they studied 28 patients, only 2 had showed radioulnar fusion without dislocation and 3 had only right sided involvement. Congenitally no other abnormality was externally detected. Family history of congenital disorders was absent. Antenatal History [as told by Mother] of drug intake in fourth month of pregnancy was noted.

Radiological examination [Figures 3 and 4]: The Ulna was represented with short ill developed upper half. Lower half of ulna and two rays were absent. His Radiohumeral and Radioulnar joints anatomically are nonexistent. No joint cavity seen at the elbow joint. Lower end of Humerus fused with the upper end of Radius and Ulna and forearm is fixed in full extension. Radius shaft was shorter than normal and curved with convexity outwards. The Carpus was shifted centrally, represented by a double row of six bones appearing to comprise the ill developed Scaphoid, Lunate, Triquetral and Pisiform and in distal row Trapizium and Trapezoid.

Figure 3: Xray of right forearm, wrist and hand showing bowed Radius, partial absence of ulna and absent ulnar rays. Figure 4: Xray of forearm with elbow, showing synostosis of elbow joint in extension.

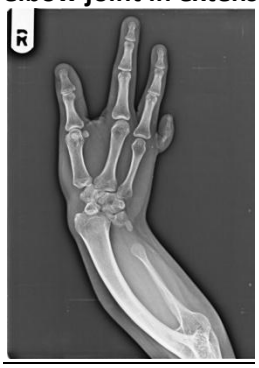

FIG 3

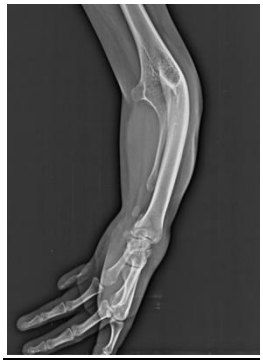

FIG 4

The two carpals, Capitate and Hamate, are absent. Three metacarpal bones present with corresponding phalanges appeared normal. Ulnar side two rays were missing. Small ill developed phalanges of the rudimentary finger were seen.

Discussion: Although chronological, tables of all the early cases of Radial, Tibial, and Fibular hemimelia are available in the literature, no Such list other than the bibliography provided by Rabaud and Hovelacque seems to have been prepared for Ulnar hemimelia.

Ulnar hemimelia was first reported in 1683 by Goller. Studies of O'Rahily[1] reveal that among the hemimelias involving one of the four bones of the third limb segment (forearm and leg); the ulnar type occur the least. It differs from the others also in that a partial deficiency is more commonly found than complete absence. However, it resembles radial, tibial, and fibular hemimelia in that it is more frequently unilateral, more commonly seen on the right side, and more often observed in the males. Hemimelia may occur as isolated anomalies or they may be associated with other malformations.

O'Rahily[1] noted the ratio of Radial to ulnar hemimelia is 18:1 and as per Abdulkadir AY[6] incidence of radial club hands between 1:55,000 to 100,000 live births and that of ulnar hemimelia occurs in about 1 in 150,000.

In 1896 only 13 cases of ulnar defect was noted. Mierzejewski (1910), found 23 cases of congenital ulnar defect. Kanavel (1932) reported 60 cases of ulnar deficiencies. O'Rahily[4] presented 65 cases in the literature up to 1950, his analysis revealed 67% of the cases were unilateral, and 69% involved right upper limb and Minoo Patel[4] revealed 70% cases are unilateral. Incidence in males was more common with a ratio of 2:1, Orphanet[6] revealed ratio 3:2. Several additional cases of ulnar hemimelia have been reported in the literature during last two decades.

Embryology –According to Abdulkadir A Y[6], most of the morphological differentiations of the limbs occur during the embryonic period. The most critical period for the development of limb anomalies is from 24-36 days of embryonic life. 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. In this case history of drug intake is noted. Minoo patel[4] mentioned that variation in longitudinal deficiencies is likely related to the timing and duration of an insult during early limb development and teratogenic insult have induced ulnar deficiencies earlier in gestation than radial deficiencies. Development of synovial joint according to Snell[7] that mesenchyme between the ends of cartilaginous bars of long bones undergoes differentiation and form capsular ligaments. Some cells in the central zone disappear and joint cavity form.

Clinical features and its etiology: 1] Absent ulna- Partial deficiency is common than complete. Abdulkadir[5] revealed the study of Ogino and Kato (1988) and according to them the critical period of ulnar deficiency 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.

2] Radial bowing – It depends upon the partial or complete absence of ulna, when ulna is partially absent then bowing is mild to moderate and When ulna is completely absent the radius may be straight. Orthopedic care center at children's hospital Boston, gave explanation on radial bowing that the improperly formed ulna may cause the wrist to deviate towards the little finger, this may cause radius to bow. While as per Minoo Patel[4] it is due to the tethering effect of the fibro-cartilagenous ulnar anlage and imbalance due to deficient lower ulnar half.

3] Synostosis with the humerus-- It is another common feature of ulnar hemimelia. Humeroradial and Humeroulnar fusion [synostosis]. Elbow is fixed either in flexion or extension. In our case elbow is fixed in

extension and mal-rotated into anterior position. Radiohumeral fusion and/ or digital syndactyly were not mentioned by O'Rahily[1]. As per Studies of O'Rahily[1]and Abdulkadir AY[5], malrotation of elbow is rarely reported. As per O'Rahily[1] that the cartilaginous skeletal elements, which are at first united by mesenchyme, joined together by fibrous tissue or by cartilage and articular cavities do not form. The absence of a radiohumeral joint (fusion) indicates the failure of cavitation of this structure. It is suggested that the lack of cavitation is an integral part of the total deficit seen in 38.5% cases of ulnar hemimelia. Works of Elhassan revealed by Abdulkadir AY[5] that, 12% of cases of ulnar hemimelia show only synostosis and 65% of cases with radio-humeral synostosis have digital anomalies.

4] Digital and carpal abnormalities – as per Minoo patel[4] digital abnormalities found in close to 90% of cases with postaxial absence of ulnar rays while O'Rahily[1] noted that three fingered hand is predominant and according to him Ulnar hemimelia accompanied by polydactyly is not unknown. Fred H Albee[8] suggested a rule that third, fourth and fifth fingers are suppressed, and in this case there is suppression of fourth and fifth fingers. According to O'Rahily[1] there is an increasing frequency of metacarpal failure as one passes from the radial to the ulnar side of the hand and the Triquetrum and Capitate often are absent, but in this case Capitate and Hamate are absent.

Radiocarpal joint has a fixed ulnar deviation and it is more useful than in case of radius absent and the presence of thumb makes it, functional hand (Fred H Albee[8]). While tethering effect of the fibrocartilaginous ulnar anlage on radial bowing, can also tether the carpus, produce limitation of wrist movement (Minoo Patel[4]).

In one of the O'Rahily[4] observation, one patient presented with oligodactyly and intrinsic muscles of hands were weak and wrist was unstable but in our case stable elbow with powerful hand grip was noted.

Management- As it is a rare anomaly, early diagnosis and careful examinations are the very important aspect of the management of cases, though the management is highly individualized. Early diagnosis [sonography] in antenatal period can reduce the occurrences of such cases.

Goal of the management is to improve function with or without the use of prosthesis, but in this case, the patient has presented after skeletal maturity and has adapted well to the anomaly for his daily living activities. Looking to the adopted functions no surgical treatment is considered for the patient. If the patient had presented in early childhood, one bone forearm reconstruction can be done which will provide cosmetic correction as well as function, in late presentations, only cosmetic correction may be required.

Association with other anomalies –According to Ballantyne, symmetrical ulnar defect is not common and associated malformations are less often present with ulnar than with radial defect **(**A R Southwood[2]). Ulnar hemimelia has been found sporadically in various anamolies and certain genetic syndromes are also associated with ulnar dysplasia—Goltz syndrome (Minoo Patel[4]).

References:

1. Charles H Frantz & Ronan O' Rahilly. Ulnar Hemimelia. J of Artificial Limbs 1971; 15: 25-35
2. A R Southwood. Partial Absence Of Ulna & Associated Structures. Journal Of Anat 1927; 61[Pt 3]: 346-351.
3. Arthur Steindler M. B. On Congenital Deformities Of The Upper Extrimity. In: Postgratuate Lectures on Orthopedic Diagnosis and Indications. volumn I. section B. USA springfield. Thomas Publisher. 1950: 243-256.
4. Patel Minoo. Ulnar Club Hand work up. URL: http://emedicine.medscape.com

/article /1243/23-overview [updated April 2010]
5. Adekunle Y. Abdulkadir. Ulnar Hemimelia With Oligodactyly- Reports Of Two Cases. Radiology Case Reports 2009 [Online]; Vol 4: No 1:240
6. Orphanet– The Portal For Rare Diseases and Orphan Drugs. Ulnar Hemimelia. URL: http://www.orpha.net/consor/cgi-bin/oc-Exp.php [October 2008]
7. Richards Snell. Clinical Embryology For Medical Students, 3rd Ed, Asian Ed, Little, Brown Company; 1983:364
8. Fred H Albee. Orthopedic and Reconstruction surgery. Industrial and civilian. Philadelphia and London. W.B. Saunders Company; 1919: 904 &905