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ULNAR INTERCALARY HEMIMELIA WITH MULTIPLE EXOSTOSES: A RARE CASE REPORT

Priyank Sahoo, Sharib Shamim, Vikas Trivedi

Department of Orthopedics

Era's Lucknow Medical College & Hospital, Sarfarazganj Lucknow, U.P., India-226003

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Address for correspondence
Dr. Priyank Sahoo
Department of Orthopedics
Era's Lucknow Medical College & Hospital, Lucknow-226003
Email: p.sahoo03@gmail.com
Contact no: +91-9199201696

ABSTRACT

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.The deficiency can be transverse, longitudinal and sometimes combination of both. This case report highlights the finding of a case of ulnar intercalary hemimelia with multiple bilateral exostoses of the knee and the rehabilitation potential for the same.

KEYWORDS: Hemimelia, Intercalary, Genetics, Multiple, Exostoses, Rare.

INTRODUCTION

Genetic defects play a major role in the transmission of anomalies. Depending on whether the anomalies are dominant or recessively inherited, the condition can manifest in off-springs. Genetic disorders, although congenital, may not be expressed until later in life. We present a case of ulnar intercalary hemimelia with multiple exostoses, which is a rare congenital defect. Ulnar hemimelia is a very rare skeletal abnormality characterized by total or partial absence of the ulna. It is reported to occur in approximately 1 per 150,000 live births. Some shortening of the forearm, radial bowing, and tendency of the hand to drift to the ulnar side of the wrist usually accompany ulnar hemimelia (1). To the best of our knowledge there is dearth of information on the combination of these anomalies in literature and their management remains challenging till date.

CASE REPORT

A 20 year old Indian unmarried girl presented to the outdoor patient department with chief complaints of deformity in her left forearm with small left upper limb and deformity in her both knees with difficulty in carrying out her daily monotonus routine. There was no history of drug ingestion other than routine antenatal drugs like folic acid and ferrous sulphate. There was also no history of TORCH(toxoplasmosis, rubella, cytomegalovirus, Herpes/Varicella virus) group of infections which is fairly common in Indian subcontinent. Also, no history of fetal alcohol, radiation, toxic substances exposure could be elicited. Evenmore metabolic causes were ruled out before hand. On examination, the left upper limb was smaller and shorter than the contra-lateral limb, showed poverty of movement but fair hand gripping and bilateral genu valgum (Fig. 1). 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 abnormalities. Radiographs (Fig. 2) showed left upper limb micromelia (2-3) and multiple exostoses around knee and (Fig. 3) left radius. She had no detectable overlying skin abnormality in her involved extremities. No significant risk factors could be assorted linked to the disease in this scenario (4).

Fig. 1: Adult Girl With Hemimelia With Bilateral Genu Vlgum And Hyper-extended Right Elbow With Ipsilateral Limb Shortening

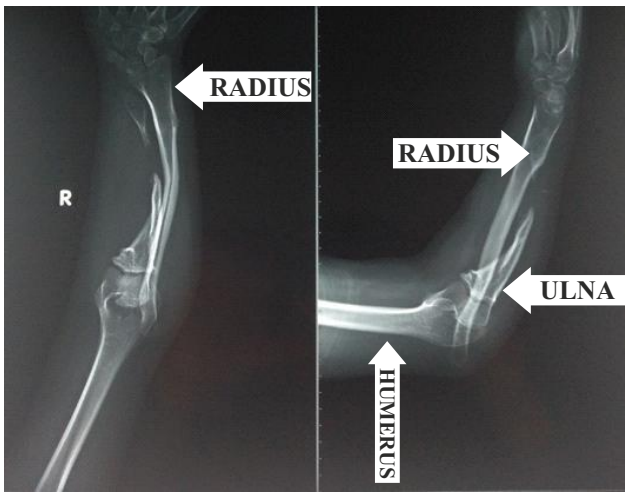

Fig. 2: Radiographs Show Right Upper Limb Ulnar Intercalary Deficiency With Radial Bowing And Radial Head Dislocation

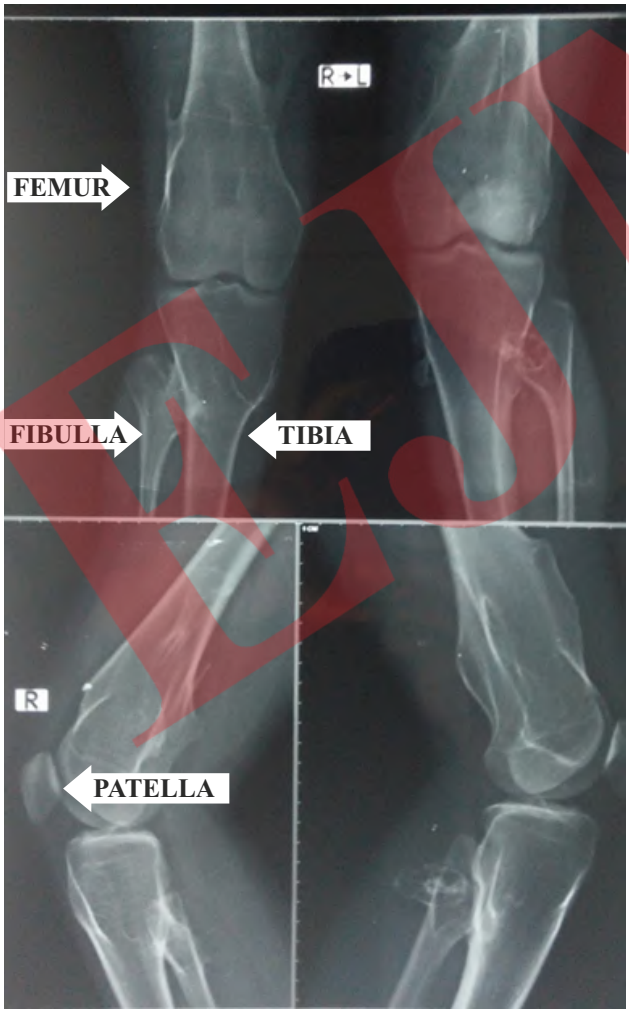

Fig. 3: Radiographs Revealing Multiple Bilateral Exostoses Affecting Both Femur, Tibia And Fibula With Valgoid Knee

DISCUSSION

Longitudinal failure of formation, of parts, or whole of a long bone in the upper limb results in congenital radial and ulnar hemimelia leading to obvious deformity. Both the radius or ulna maybe completely or partially absent, but it is rare (5). 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), thrombocytopaenia with absent radius, Fanconi anaemia (aplastic anaemia), and VACTERL syndrome. We are uncertain whether our case is a syndrome complex because of lack of information on these types of anomalies in the electronic literatures searched to the best of our knowledge. Management of such cases is dictated by age of the patient, bilaterality, levels of activity and scope of the handicap. In unilateral cases, non surgical interventions like passive stretching exercises, prosthesis, corrective casting and splinting are performed to reduce the cosmetic deformity and improve functionality of the limb. Surgery is indicated in bilateral involvement with severe handicap.[6] Non surgical management is dictated by the presentation of the child, where different prosthetic devices can be used (7-8).

No Fitting-

For children who have radio-humeral synostosis.

Opponens Post-

Children with one digit (monodigital hand) possessing good flexion power and lateral stability of the metacarpophalangeal joint were fitted to advantage

Below-elbow Prosthesis-

Modified below-elbow sockets are sometimes prescribed but the range of elbow motion is significantly lacking.

Above-elbow Prosthesis-

This is a highly satisfactory method of fitting patients with unilateral, monodigital, ulnar hemimelia. The forearm segment is acutely flexed against and parallel to the humeral shaft and then encased within the humeral socket. The elbow-locking mechanism has a lever with which the single digit controls the elbow lock and unlock mechanism.

Surgeries include elbow Z-plasty, elbow disarticulation followed by fitting the limb with a suitable prosthesis or a humeral derotation osteotomy of atleast 90 degress. To summarize, there are four approaches to treatment of the monodigital hand: op-ponens post; below-elbow prosthetic fitting; elbow-disarticulation prosthetic fitting, encasing the forearm in the humeral socket; or no fitting, which is the least recommended procedure.Rotational deformities occasionally are seen

in which there may be up to 180 degrees of medial rotation of the forearm on the humerus (9-10).

CONCLUSION

Ulnar hemimelia is a rare birth defect that causes either a partial or total absence of ulna on the affected side. Bilateral invovlement maybe seldomly seen. Some cases are mild, leading to ulnar bowing only, while severe cases may lead to complete absence of ulna along with fibrotic or absent muscle mass. Most people have shortening of forearm and limited hand-arm function. Unilateral cases have a strong predilection for the right side with female dominance. The exact cause of isolated ulnar hemimelia is not known at this time. However, most forms resulting from a syndrome do have a known genetic cause. Treatment usually includes physical therapies and surgery to help the affected arm become more functional. If your child has been diagnosed with ulnar hemimelia, talk to a doctor about all the treatment options. Support groups can provide more information and connect you with affected families (11).

CONFLICTS OF INTEREST

The authors declare that there are no conflicts of interest.

REFERENCES

1. Frantz CH, O'Rahilly R. Ulnar Hemimelia. Artificial Limbs. 1971; 15: 25-35.
2. Ogino T, Kato H. Clinical and experimental studies on ulnar ray deficiency. Handchir Mikrochir Plast Chir. 1988; 20: 330-337.
3. Elhassan BT, Biafora S, Light T. Clinical manifestations of type IV ulna longitudinal dysplasia. J Hand Surg [Am]. 2008; 33: 617.
4. 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.
5. Jain SK, Lakhtakia PK. Profile of congenital transverse deficiencies among cases of congenital orthopaedic anomalies. Journal of Orthopaedic Surgery. 2002; 10: 45-52.
6. Shafi M, Hui JHP. Common paediatric orthopaedic problems in the upper limb. Singapore Med J. 2006; 7: 654-659.
7. Johnson J, Omer GE., Jr Congenital ulnar deficiency. Natural history and therapeutic implications. Hand Clin. 1985; 1: 499-510.
8. Jain SK. A study of 200 cases of congenital limb deficiencies. Prosthet Orthop Int. 1994;18:174-179.
9. Lenz W., Der Zeitplan der menschlichen Organogenese als Massstab fur die Beurteilung teratogener Wirkungen. Fortschr. Med. 1969; 87: 520-526.
10. Layton W. M., and D. W. Hallesy, Deformity of forelimb in rats: association with high doses of acetazolamide. Science. 1965; 149: 306-308.
11. Lenz W., Zur Genese der angeborenen Hand-fehlbildungen. Chir. Plast. Reconstr. 1968; 5: 3-15.

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