Case Report

Radial Club Hand: A Neglected Case

General Paediatrics

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ABSTRACT

Radial club hand is a longitudinal deficiency along the radial side of the upper extremity. Complete absence is the most common longitudinal deficiency. It can be diagnosed on the inspection of the forearm in an X-ray. It is frequently syndromatic and so it is imperative to look for associated congenital abnormalities or syndromes by doing a thorough clinical examination and appropriate investigations. An early diagnosis and a timely operative correction give a better outcome. We report here, a 7 year old neglected case of radial club hand as an isolated anomaly.

Key Words: Radial Club Hand, Absent Thumb, Absent Radius.

INTRODUCTION

Club hand deformities are classified into two main categories, radial and ulnar. Radial club hand or radial dysplasia is an uncommon congenital anomaly. It is a longitudinal deficiency along the paraxial or the radial side of the upper extremity. It includes a wide spectrum of disorders that encompass an absent thumb, thumb hypoplasia, a thin first metacarpal and an absent radius. Ulnar club hand is much less common than radial club hand and it ranges from a mild deviation of the hand on the ulnar side of the forearm to a complete absence of the ulna. Radial club hand is frequently syndromatic [1]. Radial Club Hand usually occurs sporadically, with no known causes. An early diagnosis has a better functional outcome after reconstructive surgery.

CASE REPORT

A 7 year old male child was brought to the OPD at Narayan Medical College and Hospital, Jamuhar, Sasaram, Bihar, with complaints of cough and cold for 2 days. There was a history of a deformed left upper limb in the form of a short, curved, left forearm and an absent thumb since birth, which was ignored till date. He was a product of a non-consanguineous marriage and his perinatal history was uneventful. There was no history of blood transfusion. There was no family history of a similar deformity in the past two generations. He was developmentally normal for his age and was studying in standard two. He was anthropometrically within normal limits. The physical examination of the child revealed an atrophied and shortened left forearm as compared to the opposite normal limb and he had a single forearm bone. The movements of the left elbow flexion and extension were limited. All the distal movements including the rotatory movements of the forearm and the wrist and the fine finger movements were not possible. Only flickering movements of the fingers were possible. The thumb had not developed at all. [Table/Fig 1]. The systemic examination was normal. Evaluation of the radiographs of both the upper limbs revealed complete aplasia of the left radius and the first digit (thumb) of the left hand, including its metacarpal and the phalanges. The ulna on the affected side was shorter than that on the contra lateral side and there was radial deviation at the wrist [Table/Fig 2]. Chest X-ray, echocardiogram, haemogram including the platelet count, and ultrasound of the abdomen were normal. The child was treated symptomatically for common cold and an orthopaedic consultation was sought in order to look for available treatment options for the limb deformity correction. In view of the very late diagnosis and the limited mobility at the affected elbow joint, the parents were explained about the prognosis of reconstructive surgery.

DISCUSSION

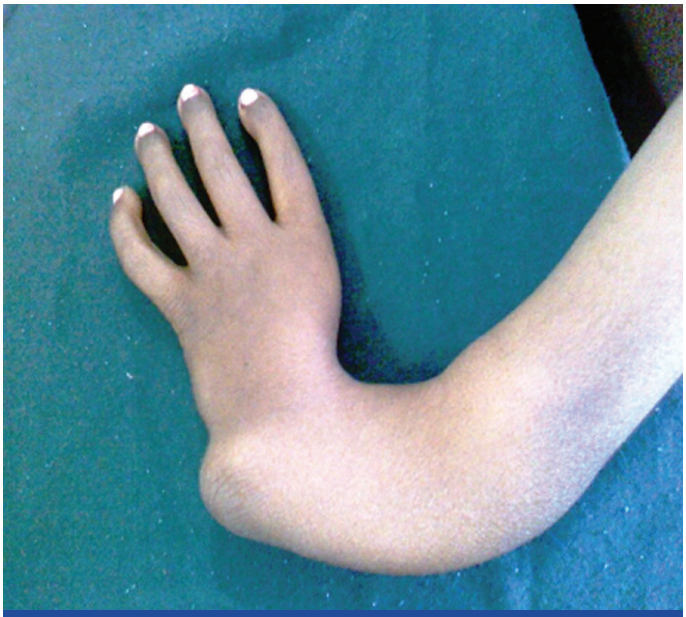
Radial club hand or radial dysplasia is an uncommon congenital anomaly. It is a longitudinal deficiency along the preaxial or the radial aspect of the upper extremity. It includes a wide spectrum of disorders that encompass an absent thumb, thumb hypoplasia, a thin first metacarpal and an absent radius [1]. The frequency of this anomaly is between 1:30000 to 1:100000 live births. The radial deficiency is bilateral in 50% of the cases and it is slightly more common in males than in females [2]. Several theories were postulated, like maternal drug exposure, compression of the uterus and vascular injury, but the current theory relates the aetiology of the radial club hand to the Apical Ectodermal Ridge (AER). AER is a thickened layer of ectoderm that directs the differentiation of the underlying mesenchymal tissue and limb formation. Removal of a portion of AER in chick embryos has produced anomalies which are similar to the radial club hand. Therefore, a defect of AER is the most probable cause of the radial club hand. The extent of the deformity is related to the degree and extent of AER absence [3]. Heikel, based on the amount of the radius which was present, classified radial dysplasia into four types, ranging from a present but defective distal radial epiphysis (Type I) to a complete absence of the radius (Type IV).

TYPE I: Mildest form with defective distal radial epiphysis.

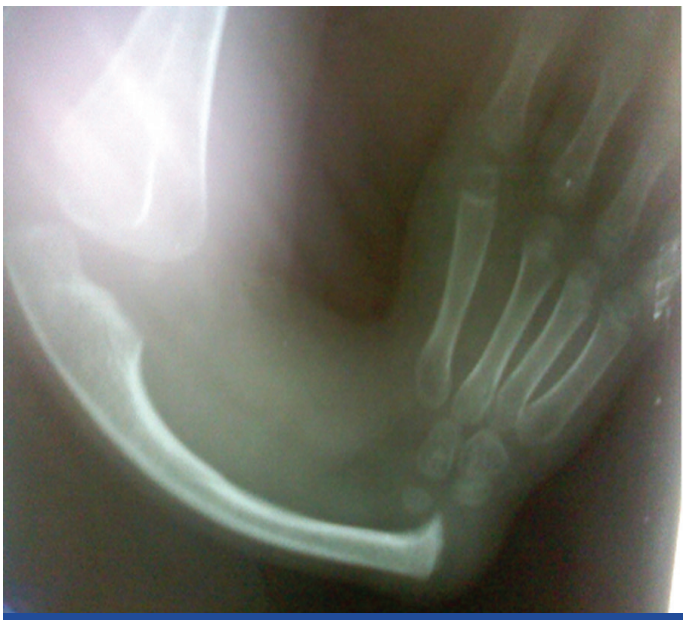
TYPE II: Involves a limited growth of the radius on both its distal and proximal sides.

TYPE III: Absence of two-thirds of the radius, most commonly the distal side.

TYPE IV: Complete absence of the radius along with a complete or a near complete absence of the thumb, which is the most common and most severe longitudinal deficiency [4].



[Table/Fig-1]: Short left forearm, absent left thumb with radial deviation of hand.



[Table/Fig-2]: X- ray left upper limb showing complete absence of radius and thumb with radial deviation at wrist.

Our case was unilateral radial club hand type IV with a complete absence of the radius, along with a complete absence of the thumb. Although the present case was diagnosed very late due to negligence by the parents, nowadays, with the availability of better medical facilities and awareness among parents, most of the radial club hands are diagnosed during the first year of life, with a better functional outcome after reconstructive surgery [5,6]. Although radial deficiency can occur in isolation, it is many times associated with other congenital malformations. Forty per cent of the patients with unilateral club hand and 27% with bilateral club hand have associated congenital anomalies involving the cardiac, genitourinary, skeletal, and the haematopoietic system [7]. Our case didn't have any associated congenital anomaly. In Radial club hand, the forearm is foreshortened, with a marked curving of the forearm, stiffness of the elbow and fingers, with the wrist being positioned in radial deviation, and the thumb being either small or absent [8]. The cardiac, genitourinary, skeletal and haematopoietic system involvement requires clinical, radiographic, echocardiogram and laboratory evaluation as appropriate. The commonly associated syndromes include:

Holt Oram syndrome: Radial dysplasia is associated with congenital heart disease (usually ASD or VSD). Abnormalities of the radius can occur in association with heart disease, but do not qualify as the Holt Oram syndrome when they are not bilateral, they lack the carpal changes and are associated with other visceral malformations and cardiac malformations which are different from intracardiac shunts, conduction disturbances or pulmonary hypertension which are characteristic of the Holt Oram syndrome.

Thrombocytopaenia Absent Radius (TAR) Syndrome: It has an autosomal recessive inheritance. The thrombocytopenia is present at birth. It is differentiated from other conditions by the presence of the thumb.

VACTERLS Association: Each letter in this syndrome's name constitutes an acronym for the defects which are involved: vertebral, anal, cardiac, tracheoesophageal, renal, limb and single umbilical artery. Babies who have been diagnosed with the VACTERLS association usually have at least three or more of these individual anomalies.

Cornelia de Lange syndrome: Children who are affected by this syndrome are usually growth retarded and they have microcephaly, classic facial features, micromelia, sensorineural hearing loss, genitourinary abnormalities and behavioural problems [9].

Fanconi's Anaemia: It is also a rare autosomal recessive disease. In infancy, there are the usual characteristic facial features (microphthalmos, strabismus and hearing defects). Pancytopaenia usually does not present until later in childhood, with the mean age of onset being 8 years. There is an increased susceptibility to malignancy, particularly leukaemia [10].

Other associations include Seckel's syndrome and an association with trisomies 13 and 18.

Our case had isolated radial club hand and didn't fit into any of the above mentioned defined syndromes. Whenever a club hand is identified, it is imperative to conduct a thorough examination and a diagnostic evaluation of the new born to delineate the associated anomalies that may suggest a syndrome. Once the birth defects have been identified, a treatment plan needs to be developed for the infant, with the gastrointestinal, renal and cardiac anomalies usually requiring early surgical management. If the patient survives these surgeries, the prognosis is usually good. The orthopaedic abnormalities can be treated individually. The non-surgical management of radial club hand involves corrective casting, bracing and physical therapy. Although surgery may be postponed for 2 to 3 years with adequate splinting, there is general consensus which favours operative correction at 3 to 6 months of age in children with inadequate radial support of the carpus. Generally, wrist reconstruction by bone graft procedures, centralization, radialization and wrist fusion is performed before thumb reconstruction by pollicization. Specific contraindications for the operative treatment include severe associated anomalies which are not compatible with long life, inadequate elbow flexion, mild deformity with adequate radial support (type I and some type II), and older patients who have accepted the deformities and have adjusted accordingly [11]. Although in our case, the child was older, with deformities in the left upper limb and contracture at the elbow joint, he accepted the deformities and the parents were not willing for operative correction. So, he was managed conservatively, but now, a better functional outcome has been documented with corrective surgery, even in older children with radial club hand [12].

CONCLUSION

Whenever a club hand is identified, it is imperative to conduct a thorough examination and evaluation of the new born to delineate the associated anomalies that may suggest a syndrome, because an early diagnosis and appropriate treatment have a better outcome.

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