# Clinical Quiz

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## A child with congenital longitudinal radial deficiency

### Clinical Presentation

A one-year-old female baby was brought to the Orthopedic Outpatient Department with complaints of outward deviation of left wrist and hand with absence of thumb since birth. The baby was born at term with unremarkable perinatal history. Examination revealed shortened left forearm and hand. The left wrist and hand were curved radially with a prominent knob at the ulnar side of wrist. There was full extension at elbow, while flexion was restricted to $100^0$. The thumb was absent (**Figure 1**). The rest of the skeletal survey was normal. Evaluation of radiograph of left forearm with wrist and hand showed hypoplastic radius with complete absence of first ray (thumb) including its metacarpal and phalanges (**Figure 2**). There was radial deviation at wrist. Routine blood investigations were normal. Sonographic and echocardiographic evaluation of the baby did not reveal any significant finding.

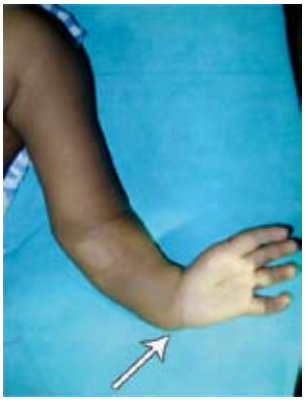

**Figure 1 -** Clinical photograph of patient showing deviation of wrist and hand to radial side with prominent knob at ulnar side of wrist (arrow head). Also note complete absence of first ray (thumb).

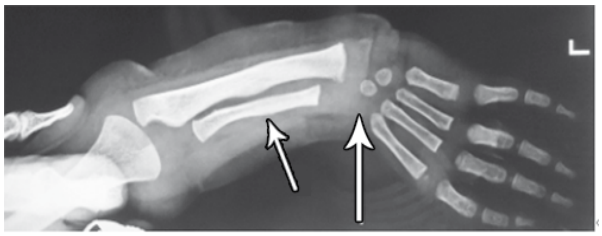

**Figure 2 -** Antero-posterior radiograph of left forearm with wrist and hand showing hypoplastic radius (short arrow) with complete absence of first ray (thumb) including its metacarpal and phalanges. Also note radial deviation at the wrist (long arrow).

# Questions

1. What is the diagnosis?

2. What other congenital anomalies are associated with this condition?

3. What is the management?

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## Answers

1. The baby has left sided congenital longitudinal radial deficiency (congenital radial club hand).

2. Although congenital longitudinal radial deficiency (congenital radial club hand) can occur in segregation, it is often associated with other congenital anomalies related to cardiac, genitourinary, skeletal, and hematopoietic system. Commonly associated syndromes include Holt Oram syndrome, thrombocytopenia absent radius syndrome, VACTERL association (vertebral anomalies, anal atresia, tracheoesophageal fistula, renal agenesis, and limb defects), and Fanconi anemia.[1-5]

3. Treatment of congenital longitudinal radial deficiency depends upon its occurrence either in isolation, or in association with other congenital anomalies. Once all the congenital anomalies are recognized, a management plan is developed first for tackling the gastrointestinal and cardiac anomalies, which usually require early surgical intervention over orthopedic deformities. If the patient survives these surgeries, the prognosis is usually good and the orthopedic deformities can be treated later on independently, either conservatively, or surgically. The conservative management of radial club hand involves corrective casting, bracing, and physical therapy, and is the definitive treatment for children with minimal deformity and stable joints.[4] Persistent wrist deviation and functionally limiting thumb deficiency requires surgical intervention. Generally, wrist realignment is performed first by centralization, or radialization of wrist before thumb reconstruction by pollicization. The optimal time is between 6 and 12 months of age especially if it is intended later to pollicize the index finger.[4]

## Discussion

Radial longitudinal deficiency encompasses a spectrum of dysplasias and hypoplasias involving the thumb, wrist, and forearm. The severity of deformity ranges from mild thumb hypoplasia to complete absence of the radius.[4] It is an uncommon congenital anomaly. The most common presentation is complete absence of radius.[5] Radial longitudinal deficiency usually occurs sporadically with no identified cause. In our case, the patient had hypoplasia involving the proximal and distal end of radius with complete absence of first ray (thumb). Bayne and Klug[6] classified radial deficiency into 4 types, ranging from a defective distal radial epiphysis (type I) to complete absence of the radius (type IV).[5] Type IV is the most common and most severe longitudinal deficiency. Type I is mildest form with defective distal radial physis. Type II involves restricted growth of the proximal and distal radial physis. Type III - partial absence of radius, most commonly the distal part and type IV, complete absence of the radius. Our case was Bayne and Klug[6] type II with hypoplastic proximal and distal ends of radius. Congenital longitudinal radial deficiency is commonly associated with other anomalies, as the development of several organ systems coincides with development of the upper limb buds. In our case, congenital longitudinal radial deficiency was detected in isolation. The goals of treatment of congenital longitudinal radial deficiency is to optimize limb length discrepancy, correction, and maintenance of wrist deformity, preservation of wrist mobility, and longitudinal growth of ulna, and to attain an acceptable cosmetic result,[4] either by conservative, or surgical approach.

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