

# Radial Club Hand — A Case Report and Review of Literature

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## Abstract

Radial club hand refers to deficiency along the preaxial side of the extremity. Complete absence is the most common longitudinal deficiency. It is diagnosed after inspection of the forearm and an X-ray. It is however imperative to look for associated congenital abnormalities or syndromes regarding the cardiovascular system, renal system, vertebral column, blood cells and digestive system of the child by appropriate investigations. This article presents a case of radial club hand, highlighting the approach to diagnosis with multisystem evaluation and reviews the management of such patients.

## Introduction

Radial Club Hand is an uncommon congenital anomaly. Although the complete absence is the most common presentation, it ranges from hypoplasia of the thumb to various degree of radial hypoplasia. Radial Club Hand usually occurs sporadically with no known cause. There is no known genetic link except when it is associated with other congenital anomalies or syndromes with a known genetic component.

## Case Report

A six day old neonate was referred for evaluation of congenital deformity of right upper limb. The baby was born at term and her perinatal history was unremarkable. The mother of the baby was a 38 year old elderly primipara and diabetic but denied any other relevant obstetric history. Physical examination of the baby revealed imperforate anus, along with deformity of right forearm which appeared shorter than contralateral side with radial deviation at the wrist and absent thumb. Routine blood investigations were normal. Evaluation of the radiographs of both upper limbs revealed complete aplasia of radius and first digit (thumb) including its metacarpal and phalanges, the ulna on the affected side was shorter

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than contralateral side and there was radial deviation at the wrist. X-ray of lumbar spine revealed L3 hemivertebra. Sonographic and echocardiographic evaluation of the neonate did not reveal any significant finding.

## Discussion

Radial club hand or radial dysplasia a rather uncommon congenital anomaly refers to deficiency along the preaxial or radial side of the extremity. Its incidence varies from 1 in 30,000 to 1 in 100,000 live births.<sup>1</sup> It ranges from hypoplasia of the thumb to various degree of radial hypoplasia.<sup>2</sup> Bayne and Klug classified radial deficiency into four types, ranging from a present but defective distal radial epiphysis (Type I) to complete absence of the radius (Type IV).<sup>3</sup> Complete absence (Type IV) is the most common and most severe longitudinal deficiency.

TYPE I: Mildest form with defective distal radial epiphysis.

TYPE II: Involves 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: This type of Radial club hand is the most common and the most severe and involves complete absence of the radius along with complete or near complete absence of

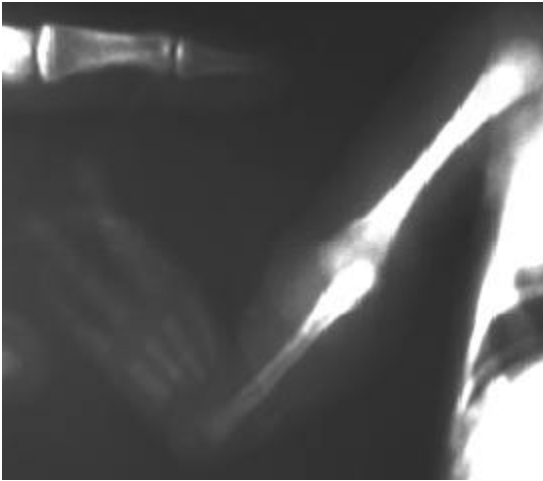


Fig. 1 :X- ray right upper limb showing complete absence of radius and thumb with radial deviation at wrist.

the thumb.

Our case was Type IV category of radial club hand with complete absence of radius along with complete absence of the thumb. Radial club hand usually occurs sporadically with no known cause. Although radial deficiency can occur in isolation it is many times associated with other congenital malformations. Forty per cent of patients with unilateral club hand and 27% with bilateral club hand have associated congenital anomalies.<sup>4</sup> Cardiac, genitourinary, skeletal and haematopoietic system involvement requires clinical, radiographic and laboratory evaluation as appropriate. Commonly associated syndromes include:

1. *Holt Oram syndrome*: Radial dysplasia 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 Holt Oram syndrome when they are not bilateral, lack the carpal changes and are associated with other visceral malformations and cardiac malformations different from intracardiac shunts, conduction disturbances or

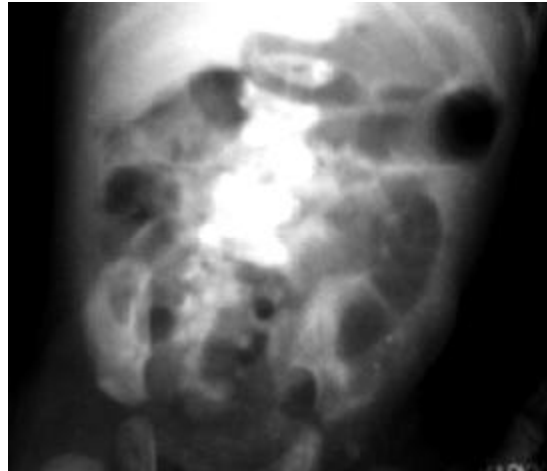


Fig. 2 :X-ray lumbar spine showing L3 hemivertebra.

pulmonary hypertension characteristic of Holt Oram syndrome.<sup>5</sup>

2. *Thrombocytopenia 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.

3. *VACTERLS Association*: Each letter in this syndrome's name constitute an acronym for the defects involved: vertebral, anal, cardiac, tracheoesophageal, renal, limb and single umbilical artery. The cause is unknown but it is a non random association whose simultaneous occurrence by chance is unlikely.<sup>6</sup> Babies who have been diagnosed with VACTERLS association usually have at least three or more of these individual anomalies.

4. *Fanconi Anaemia*: It is also a rare autosomal recessive disease. In infancy there are usual characteristic facial features (microphthalmos, strabismus, hearing defects).<sup>7</sup> Pancytopenia usually does not present until later in childhood with the mean age of onset being 8 years. There is increased susceptibility to malignancy particularly leukaemia.

5. *Cornelia de Lange syndrome*: Children

affected with this syndrome are usually growth retarded, have microcephaly, classic facial features, micromelia, sensorineural hearing loss, genitourinary abnormalities and behavioural problems.

6. Other associations include Seckels syndrome and association with Trisomy 13 and 18. The baby in our case was thus diagnosed as having VACTERLS Association (Radial club hand, imperforate anus and vertebral anomaly). The most obvious physical finding at birth in this association is the radial ray defect. Between 5 – 10% of radial club hands are associated with VACTERLS, thus underscoring the need of multisystem evaluation in babies born with radial club hand.

Although no specific genetic or chromosome problem has been identified with VACTERLS Association, it can occasionally be seen with trisomy 18 and is more frequently seen in babies of diabetic mothers. The vertebral defects usually consist of hypoplastic or hemivertebra. Thoracic anomalies are worse in those with tracheo-oesophageal fistula and lumbar anomalies are more common in those who have imperforate anus. MR Study of the spine is recommended for detection of any occult intraspinal pathology and in those who require operative management for their scoliosis. Ventricular septal defect is the most common cardiac problem. Upto 35% of patients with VACTERLS Association have a single umbilical artery which can often be associated with kidney or urologic problems. The limb anomalies range from a hypoplastic thumb to radial club hand, which may be unilateral or bilateral, bilateral defects being always asymmetric.<sup>8</sup> Lower limbs are affected only occasionally.

Once the birth defects have been identified, a treatment plan needs to be developed for the infant with the gastrointestinal 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. Persistent wrist radial deviation contracture and functionally limiting thumb deficiency requires surgical intervention. Generally wrist reconstruction by bone graft procedures, centralization, radialization and wrist fusion<sup>9</sup> is performed before thumb reconstruction by pollicization.

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