

## Case Report

# RADIAL RAY SYNDROME-A CASE REPORT

**\*Ramesh G.<sup>1</sup>, Sudha R.<sup>1</sup>, Vimala<sup>1</sup>, Santhosh<sup>2</sup> and Siddharth<sup>1</sup>**

<sup>1</sup>Department of Obstetrics and Gynecology, MVJ Medical College and Research Hospital,  
Hoskote, Bangalore, India

<sup>2</sup>Department of Paediatrics, MVJ Medical College and Research Hospital,  
Hoskote, Bangalore, India

*\*Author for Correspondence*

## ABSTRACT

Radial ray defect is a rare congenital defect that may be isolated or associated with other anomalies. Some of the well-known combinations are Fanconi' syndrome and Holt – Oram syndrome. We report a rare case of radial ray syndrome with no association with other anomalies.

**Key Words:** Radial Ray, Anomaly

## INTRODUCTION

Radial ray defect is a rare congenital defect that may be isolated or associated with other anomalies .some of the well-known combinations are Fanconi' syndrome and Holt – Oram syndrome<sup>1</sup>.

The radial ray consists of one solid bone, the radius, and a segmented portion composed of the scaphoid, the trapezium, the first metacarpal, and the two phalanges of the thumb. All these bones are absent in total terminal defect of the ray. In partial terminal deficiency, the first metacarpal and two phalanges of the thumb and sometimes varying portions of the distal radius may be missing.

## CASES

Mrs. X, a 19 yr. old Primigravida presented in the outpatient department with H/o 9 months of amenorrhoea with c/o labour pain and h/o leaking p/v since 4 hrs . Her antenatal period was uneventful with normal investigations.

On examination, patient was moderately built, a febrile with mild pallor. Uterus was term size, intermittent regular contractions, with cephalic presentation. FHS heard. On P/V examination cervix was fully effaced and fully dilated, Vertex at +1 station and membranes absent.

FTND with RMLE was done and a live female baby was delivered with birth wt. of 2.1 kg. Baby cried immediately after birth.



**Figure 2:** Shows X-ray of the baby depicting absent radius and absent left thumbs.



**Figure 1:** Shows photograph of the baby depicting absent thumb in the left hand.

## Case Report

On examining the baby we found a deformity in the upper limbs. Right hand had a rudimentary finger-like cystic sac (? polydactyl) and left hand showed the presence of amniotic band and an absent thumb. The left forearm appeared to fold upon itself and on palpating the forearm bones were not felt. Except for the absence of thumb and radius, the baby was otherwise healthy. Figure 1 Shows photograph of the baby depicting absent thumb in the left hand. X-ray examination of baby's whole body revealed absent thumb and absent radius in the left upper limb. Figure 2 Shows X ray of the baby.

Ultrasound examination for ruling out internal organ malformations was reported to be normal. All blood counts (including platelet count) and other biochemical investigations were normal.

Patient gives no history of consumption of any medication or h/o any fever etc. during her antenatal period. Postnatal period was uneventful.

## DISCUSSION

Radial ray anomalies comprise of a large spectrum of anomalies which ranges from partial (Radial Hypoplasia) to a complete (Radial Aplasia) deficiency of radius +/-bones of the thumb. They can be associated with many syndromes; Aase syndrome, amniotic band syndrome, comelia de Lange syndrome. Holt-Oram syndrome, fanconi anemia, Tar syndrome, Rothmund Thomson syndrome<sup>2</sup>. Antenatal ultrasound may show absent or hypoplasia of radius with hand often in medial rotation. 1 in 100000 children are born with radial ray anomalies. They are more common in males and in Caucasians. This condition affects both arms in 38% - 50% of children. It results from injury to the developing arm during fourth to seventh week of pregnancy. In some cases this can be caused by exposure to factors in the environment including compression, inflammation, nutritional deficiency and chemical and drug exposure. Radial ray anomalies can be classified into four main subtypes depending on the extent of severity.

**Type I:** Radius is **slightly** (> 2 mm) short and the hand to bends sideways at the wrist (often associated with a hypoplastic thumb); proximal radius usually unaffected.

**Type II:** The radius bone is **very short** and the ulna curves sideways and supports the wrist poorly.

**Type III:** Partial absence of radius.

**Type IV:** Complete absence of radius.

Usually no specific deformity cause is found. Plastic surgeon usually does surgery to correct and to achieve the best possible functional use of the hand. Surgery may also be recommended for hypoplastic thumbs.

Kutsal et al has reported a case of radial ray defect with vascular and vertebral anomalies in Archives of Orthopaedic and Trauma Surgery (1989). It was a radial ray defect along with high origin of radial and ulnar arteries, and anomalies of thoracic vertebrae. The case was regarded as a thoracic outlet syndrome due to the first rib abnormality and was treated surgically.

Cox et al reported 34 cases with defects of the radial ray, 24 individuals had additional clinical manifestations. A firm syndromic diagnosis could be reached in 17 persons (TAR syndrome 4, Holt-Oram syndrome 8, Fanconi anaemia 2, VATER association 2 and Radial ray-choanal atresia 1). In the remainder, no specific diagnosis could be established. The heterogeneity of radial ray syndromes has important implications for prognostication and genetic counseling *et al.*, (1989).

Sevim Balci et al reported a case of omphalocele with absent thumb .they suggested that the association of the omphalocele and radial ray dysplasia may not be coincidental. They may share a common etiology. A more fitting terminology for this association was named as omphalocele-radial ray defects (ORRD) complex Sevim *et al.*, (2005).

The challenge of radial ray anomalies is to combine clinical and ultrasound expertise with input from clinical genetics, ultrasound and molecular testing. Kennelly, M. M and Moran P has proposed a clinical algorithm which encourages targeted sonography including 3D views for subtle face, ear and hand anomalies, providing a useful tool to diagnose the underlying condition, crucial for appropriate obstetric management and prognosticating for future pregnancies Kennelly and Moran (2007).

### **Case Report**

#### **CONCLUSION**

Radial ray defect is a rare congenital anomaly which may be isolated or can occur in association with well-known syndromes like Fanconi syndrome, Holt – Oram syndrome, TAR syndrome. Newborn should be evaluated with complete blood count, X-ray of the limb, Echocardiography and ultrasound of the abdomen to rule out other associated anomalies.

#### **REFERENCES**

- Archives of Orthopaedic and Trauma Surgery (1989).** 108 333-335.
- Mehollin Ray AR et al., (2008).** Radiographic abnormalities in Rothmund Thomson syndrome and genotype, phenotype correlation with relqly mutation states. *American Journal of Roentgenology* **190**(2) 62.
- Cox H, Viljoen D and Versfeld G (1989).** Beighton P Radial ray defects and associated anomalies. *Clin Genet.* **35**(5) 322-330.
- Sevim Balci, Gürsel Leblebicioglu and İnci Yigitkanlı (2005).** A new case of omphalocele with absence of thumb. *Turk Journal Pediatr* **47** 95-97.
- Kennelly MM and Moran P (2007).** A clinical algorithm of prenatal diagnosis of Radial Ray Defects with two and three dimensional ultrasound. *Prenatal Diagnosis* **27** 730–737.