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Case Report

PRENATAL FATAL RADIAL RAY MALFORMATION WITH CLEFT LIP AND PALATE – A CASE REPORT

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ABSTRACT
A lethal variant of Radial Ray Malformation with the following features is being presented i.e., bilateral radial ray manifestation, (Radial aplasia) complete absence of radius of left forearm leading to hemimelia, bilateral absence of pollex, and bilateral pulmonary hypoplasia. Cleft lip and palate existed as associated features. The radial ray malformations can occur as a single defect or in some cases they exist associated with other systemic deformities. The hypoplasia of lungs , is responsible for the fatal form of Radial Ray Malformation.

Key Words: *Radial Ray, Radial Aplasia, Hemimelia, Pollex, Cleft Lip, Cleft Palate And Hypoplasia.*

INTRODUCTION
Knowledge of the limb defects such as absence of fingers brought out by Aristotle long ago, however radial ray defects were not brought to the fore. Radial Ray malformation is a rare congenital abnormality with an incidence of one in Thirty thousand live births James(2004) characterised by fixed abnormal medial rotation of both the hands at wrist joint, aplasia or hypoplasia of radius with associations as VACTERAL association ,Thrombocytopenia, cardio vascular anomalies as in Holt-Oram syndrome (heart-hand syndrome) (Woodward *et al.,* 2004). In the present case right radial aplasia, bilateral fixed abnormal medial rotation of both the hands at wrist joint other associated features including cleft lip and cleft palate and pulmonary hypoplasia were observed. Such cases are very rare and if occur are fatal.

CASE
A 27 year old female with a five year marital life, G2P1 with no history of consanguity and no family history of congenital anomalies, delivered a male still born foetus of 30weeks gestation with gross anatomical abnormalities at MIMS general Hospital.

Foetal Autopsy - Findings

External Features:

Complete Cleft lip and Cleft palate on right side.
Flattened nasal bridge (saddle shaped).
Bilateral Radial Ray malformation.
Bilateral absence of Pollex.
Lower limbs did not show any external variations (Fig- 1.a).
No abnormalities in the ears.
Anal atresia is not found.

Foetogram revealed

Absence of Radius on the right forearm (Radial aplasia) only.
Bilateral fixed and medially rotated hands at wrist joint.
Missing pollex of Left hand with normal radius and ulna.
Increased intervertibral disc spaces (Fig- 1.b).

Internal Features:

Bilateral Hypoplastic Lungs.
Defects in the internal structure of Heart could not be elicited.

DISCUSSION

Yuranga (2010) classified Radial Ray malformation into four types i.e., Type I : where radius is slightly (> 2 mm) short and the hand bends sideways at the wrist (often associated with a hypoplastic thumb) ; proximal radius usually unaffected .Type II : radius is very short and the ulna curves sideways to support the wrist .Type III : partial absence of radius . Type IV: complete absence of radius. Absence of thumb is also a feature. The present case falls into Type IV radial ray as complete absence of radius on the right side with abnormally fixed and medially rotated left hand along with normal development of radius in the forearm.

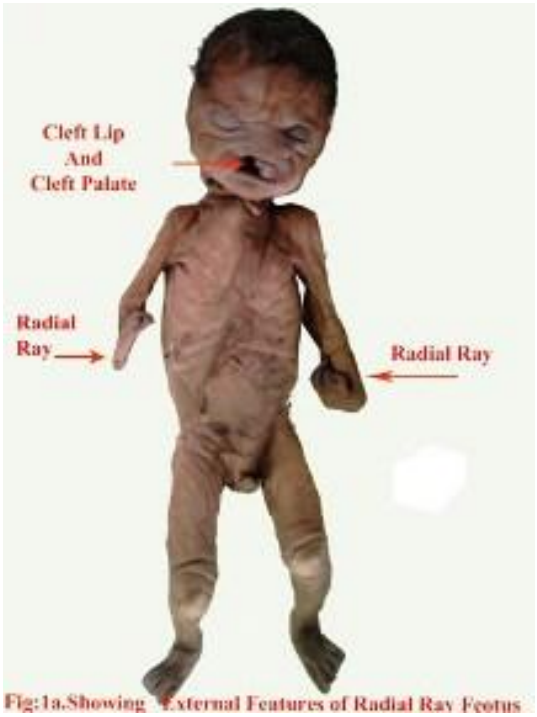

Fig:1a.Showing External Features of Radial Ray Foetus

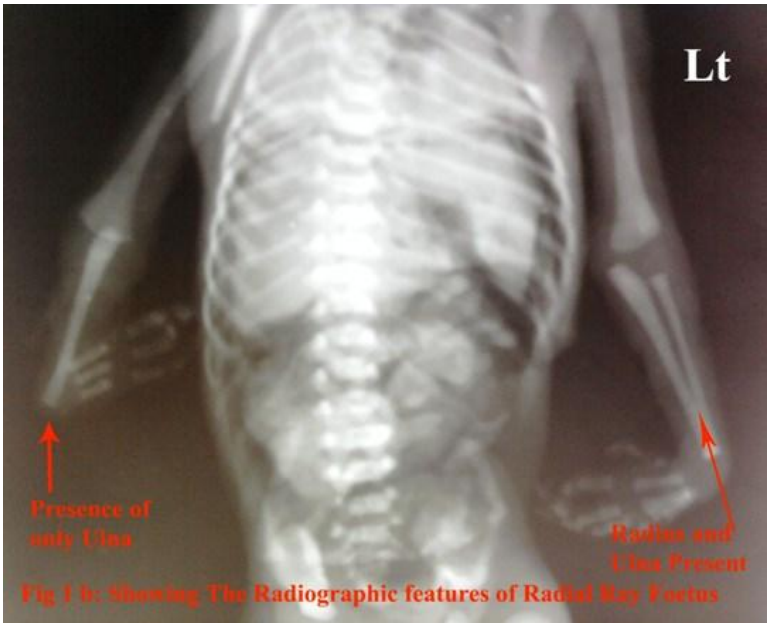

Fig 1 b: Showing The Radiographic features of Radial Ray Foetus

The present case is differentiated from TAR (Thrombocytopenia Absent Radius) syndrome as in TAR even though radius is absent and thumb is always present (Radswiki 2011). The present case is also differentiated from VACTERAL syndrome and Holt- Oram syndrome due to absence of features like tracheoesophageal fistula, anal atresia, renal anomalies and visible cardiovascular anomalies. This is not a case of Fanconi anemia as partial reduction of radius will be there in Fanconi and the present case showed complete aplasia of the radius. Association of cleft lip and palate along with radial ray has not been reported in literature.

In Trisomy 13 along with cleft lip and cleft palate, cardiovascular anomalies, limb deformities have been reported (Cowen 1979); which are not the associates of present case of radial ray anomaly.

Conclusion

Evolution of techniques such as Ilizarov technique (which is an orthopaedic procedure for lengthening the long bones by the induction of new bone formation between the opposing bone surfaces which resulted from controlled pulling apart) (Ali *et al.*, 2008) and maxillofacial surgeries can bring the life style of patients with non fatal congenital skeletal malformation closure to normalcy. It is of immense importance to be able to assess the non fatal forms prenatally. In the present case the hypoplasia of the lungs is the only feature which made it fatal. The knowledge of the existence of such anomalies helps to arrive at an appropriate diagnosis to influence the decision of continuing pregnancy by the mother so as to prevent the furtherance of complications.

REFERENCES

Ali Al Kaissi, Klaus Klaushofer, Alexander Kerbs and Franz Grill (2008). A Novel malformation complex of bilateral and symmetric preaxial radial ray-thumb aplasia and lower limb defects with

minimal facial dysmorphic features: a case report and literature review. *Cases Journal* **1**:271 [Avalable at http://www.casesjournal.com/content/1/1/271].

Cowen J M, S Walker, and F Harris (1979). Trisomy 13 and extended survival. *Journal of Medical Genetics.* 16(2) 155–157.

James Wynbrandt and Mark D Ludman (2004). The Encyclopedia Of Genetic Disorders and Birth defects 2nd Edition Published by Viva Books as an arrangement with Facts on File. Inc New York. 2004 p No: 276.

Radswiki (2011). TAR Syndrome. ID No: 14658. Available at www.Radiopaedia.org.htm

Woodward, Kennedy, Sohaey, Byrne, Oh and Puchalski (2004). Diagnostic Imaging Obstertrics 1st Edition Published by AMIRSYS /ELSEVIER Utah, 10th section p No: 38- 40.

Yuranga Weerakkody (2010). Radial Ray Anamoly Article ID 10802 Available www. Radiopaedia.org.htm