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Radial Ray Defect

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Author's contribution

The sole author designed, analyzed and interpreted and prepared the manuscript.

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Case Report

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ABSTRACT

A 24 days old male neonate presented to the University of Gondar Hospital with deformity at the right wrist joint since birth. Physical findings were deformity of the wrist joint with lateral deviation of fingers and absent thumb. Plain radiographs of the wrist joint showed absent radius and bones of the thumb which suggest the diagnosis of radial ray defect. Radial ray defect is a rare congenital defect that may be isolated or associated with other anomalies. This is a rare case of radial ray defect which has no association with other anomalies.

Keywords: Radial ray; defect.

1. INTRODUCTION

1.1 Radial Ray Defect

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 pancytopenia syndrome, Holt – Oram syndrome, Thrombocytopenia-absent radius (TAR) syndrome, VACTERL syndrome [1,2].

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Current theory relates the etiology of radial ray defect to the apical ectodermal ridge (AER). This structure is a thickened layer of ectoderm that directs differentiation of the underlying mesenchymal tissue and limb formation. Removal of a portion of the AER in chick embryos has produced anomalies similar to radial clubhand [3]. Therefore, a defect of the AER is the most probable cause of radial ray defect, with the extent of deformity related to the degree and extent of the AER absence [4].

The radial ray consists of one solid bone, the radius, and a segmented portion composed of the scaphoid, trapezium, first metacarpal, and two phalanges of the thumb. Radial ray anomalies comprise of a large spectrum of anomalies which ranges from partial (Radial Hypoplasia) to a complete (Radial Aplasia) deficiency of radius with the presence or absence of bones of the thumb.

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.

2. CASE REPORT

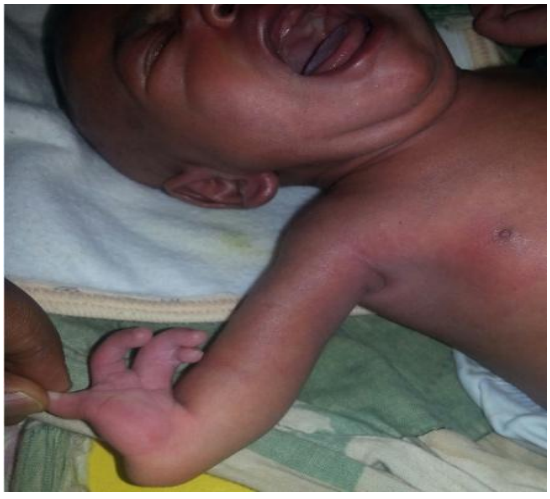
A 24 days old male neonate presented with deformity at the right wrist joint since birth. The delivery was at Kola duba health centre which is spontaneous vaginal delivery. The mother lives in rural areas and she is Para 2 lady and no similar illness in the family. The duration of the pregnancy is not clearly known by the mother but is claimed to be amenhoric for 9 months. The mother had no maternal illness during pregnancy and no medication taken at the time of pregnancy. She has no antenatal care (ANC) follow up during pregnancy.

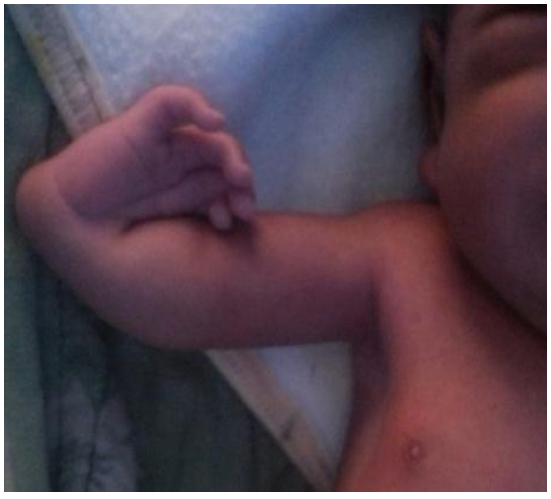
On physical examination, there is a deformity at the right wrist joint with lateral angulation of the fingers. Only 4 fingers are seen with the absence of the thumb. No other pertinent physical finding seen (See Figs. 1,2).

Complete blood count (CBC) is normal.

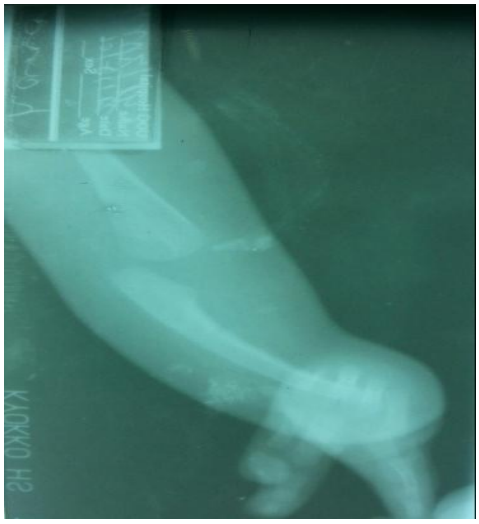
Forearm radiograph shows absent radius, deformity of the hand with radial angulation, absent metacarpal and phalangeal bones of the right thumb (See Figs. 3,4).

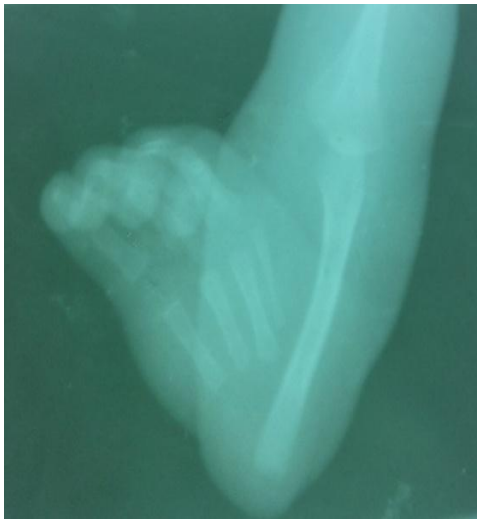
Abdominal ultrasound and echocardiography were also done and no any abnormality seen.





Figs. 1 and 2. Photograph of a baby with the wrist deformity and absent thumb





Figs. 3 and 4. AP and lateral right forearm radiograph of the baby with absent radius and bones of the thumb

3. DISCUSSION

Radial ray defects are rare congenital anomalies. The presence of bilateral radial ray defects increases the likelihood of a syndromic diagnosis. Syndromes with this type of upper limb defect are Fanconi pancytopenia syndrome, Holt-Oram syndrome, VATERR association and TAR syndrome [5]. 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 [6]. 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.

Fanconi pancytopenia syndrome, an autosomal recessive condition was first described in 1927 [7]. This syndrome consists of radial hypoplasia, hyper pigmentation and pancytopenia. Fanconi's was ruled out as the diagnosis in our case because the complete blood count is normal and no skin manifestation in the child. Chromosomal diagnosis of Fanconi pancytopenia is through induction of chromosomal breakage by diepoxybutane (DEB) but it is not available in our hospital.

Holt-Oram syndrome was first reported in 1960 and consists of upper limb defects and cardiovascular anomalies. It is an autosomal dominant condition but with variable penetrance [8]. The upper limb defects may occur with different degrees of severity. The thumbs may be absent, hypoplastic, triphalangeal or bifid. The first metacarpal or radius may be absent or hypoplastic. There may also be defects affecting the ulna, humerus, clavicle, scapula or sternum. The cardiovascular anomalies may be atrial septal defects, ventricular septal defects, conduction defects or hypoplasia of distal bloodvessels. In our case, although the upper limb defects were present, there was no cardiovascular abnormality.

VATERR association is not a syndrome. Association describes the phenomenon where some malformations present collectively more frequently than you would expect by chance [9]. The acronym VATERR stands for Vertebral defects, Anal atresia, TE (tracheo-oesophageal) fistula with oesophageal atresia, Radial dysplasia and Renal dysplasia. Cardiac defects, single umbilical artery and growth deficiency have also been reported. Radial dysplasia occurs in 65% of reported cases and include hypoplasia of the thumb or radius, preaxial polydactyly or syndactly. In our case, there was no evidence of vertebral or renal abnormalities on ultrasound scan. It was not possible to exclude anal atresia or a tracheoesophageal fistula on scan but there was no feeding and chest problems after birth. The only feature of VATERR in our case was the radial dysplasia so the other features of VATERR association were absent and therefore make VATERR unlikely to be the cause.

TAR syndrome comprising radial aplasia and thrombocytopenia was first described in 1956 [10]. It is a microdeletion syndrome. The defects occur in both upper and lower limbs. There may be bilateral absence of the radius, abnormalities of the ulna such as hypoplasia, bilateral absence or unilateral absence; abnormal humerus, abnormal shoulder joint but the thumbs are always present. Forty percent of the live births reported have died as infants from haemorrhage. TAR syndrome has also been ruled out as the diagnosis, because the neonate did not have lower limb defects even though lower limb defects are not present in all reported cases of TAR syndrome. The other difference was that TAR syndrome babies always have thumbs. In our case there was an absent right thumb. Most importantly the TAR syndrome babies always have bilateral absence of the radius [11] which was not the presentation in this case.

Correction of radial ray defect requires a combination of non-operative and operative management that begins shortly after birth. Instruction in passive stretching of the taut radial structures is provided at the initial visit. This stretching is performed at each diaper change and at bedtime. Splint fabrication is difficult in the newborn, especially with a shortened forearm. Therefore, splint use is delayed until the forearm is long enough to accommodate a splint. Serial casting can also be used to gradually stretch the tight radial structures.

Centralization of the wrist on the ulna is the standard treatment to correct radial deviation. This procedure is performed in patients aged approximately 1 year. Surgery at this time allows improvement in forearm length and provides a foundation for the development of motor function within the hand.

Finally our case couldn't fit to one of the above syndromes so it is a case of isolated radial ray defect which is a very rare congenital anomaly. There is no a single case report seen in literatures before in Ethiopia.

4. CONCLUSION

Radial ray defect is a rare congenital anomaly which may be isolated or can occur in association with other anomalies. Newborn with radial ray defect should be evaluated with complete blood count, chromosomal analysis, X-ray of the limb, Echocardiography and ultrasound of the abdomen to rule out other associated anomalies.

CONSENT

Written informed consent was obtained from the patient for publication of this case report and accompanying images.

ETHICAL APPROVAL

All authors hereby declare that all experiments have been examined and approved by the Appropriate ethics committee and have therefore been performed in accordance with the Ethical standards laid down in the 1964 Declaration of Helsinki.

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COMPETING INTERESTS

Author has declared that no competing interests exist.

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