VOLUME-8, ISSUE-3, MARCH-2019 • PRINT ISSN No 2277 - 8160

Original Research Paper

Neonatology

CONGENITAL LONGITUDINAL RADIAL DEFICIENCY-A CASE REPORT

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ABSTRACT

BACKGROUND-The spectrum of radial longitudinal deficiency is extensive which could be isolated or associated with other anomalies.

CASE CHARACTERISTICS- A male neonate presented with deformity of right upper limb along with hypoplasia of thumb , X-ray of right forearm showed complete absence of radius bone along with hypoplastic thumb.

OBSERVATION- Isolated radial deficiency is usually a rare entity and thorough examination should be done to rule out other anomalies.

MESSAGE- Centralization of wrist on ulna is the standard treatment usually done by 12 months of age.

KEYWORDS : radial longitudinal deficiency, neonate, anomalies, centralization of wrist.

INTRODUCTION:

Congenital longitudinal radial deficiency (CLRD) is a relatively rare congenital anomaly characterized by variable degree of deficiency along the radial (or preaxial) side of the limb [1].Half of the radial ray anomalies are inherited in a mendelian pattern and remaining half sporadically[2].It results from mutations in the SALL4 gene [3].This process was formerly referred to as radial club hand [4].Frequency of this anomaly is between 1:55000 to 1:100000 live births[5].The incidence of all radial ray-deficient limbs, including patients with hypoplastic thumbs alone, is approximately 1 in 30,000 [1].The prevalence has been reported to be slightly higher in boys than in girls and right sided involvement being more[6].

CASE REPORT

A male neonate was born to a 23 years old primi para mother through elective cesarean section in view of cephalopelvic disproportion. Antenatal history was insignificant. The baby presented with abnormally short right forearm. Physical examination of the baby showed deformity of right forearm, with radial deviation at wrist and hypoplasia of the thumb (figure 1A,B). Routine blood investigations were within normal limits. Plain radiographs of the right forearm with hand were taken and it showed complete absence of radius and hypoplastic thumb (figure 2A). X-ray of chest did not show any abnormality (figure2B). USG abdomen and echocardiography of the baby did not reveal any significant finding.

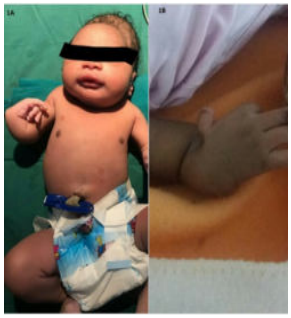

Figure1A – Image of the male neonate showing deformity of right forearm and radial deviation of wrist. **1B**- Hypoplasia of thumb.

DISCUSSION-

Radial longitudinal deficiency has been classified into 4 types by Bayne and Klug based on radiographic severity of the radial ray deficiency[4].Type I is the mildest form. There is a short radius and minor radial deviation of hand. TypeII has hypoplastic radius with abnormal growth at proximal and distal ends and moderate radial deviation of hand. In Type III there is partial absence of radius and severe radial deviation of hand. In Type IV there is complete absence of the radius.This is the most common type of all[4].Most often Radial longitudinal deficiency is associated with various other anomalies like Cardiovascular include Holt-Oram syndrome, VACTERAL association,

Ventriculoradial dysplasia[7],Genitourinary includes ectopic kidney, hypoplastic kidney , horseshoe kidney[4],Skeletal conditions associated with it are Klippel-Feil syndrome, Keutel syndrome (costovertebral dysplasia-humeroradial synostosis) ,VACTERAL [4],Hematologic conditions are Aase-Smith syndrome, Fanconi anemia, TAR syndrome-Thrombocytopenia-Absent radius[7],Chromosomal causes like trisomy of 13, 17, 18 [7], Teratogenic causes like Thalidomide embryopathy[7].Isolated congenital radial longitudinal dysplasia is rare. In the present case the baby was investigated for the above mentioned syndromes. X-ray of chest did not show any hemivertebrae or rib anomalies (figure2B).Blood investigations , USG abdomen and 2-D echo did not show any significant abnormality. Isolated radial longitudinal deficiency is very rare and its incidence also being rare as mentioned above. Whenever a club hand is identified it is important to conduct a thorough examination of fetus and new born to delineate associated anomalies that may suggest a syndrome. Generally speaking this is a useful sign in differentiating isolated form from that associated with syndromes.

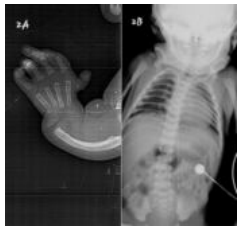

Figure 2

2A-X-ray of the right forearm shows typeIV congenital longitudinal radial deficiency with rudimentary 1st metacarpal and absent proximal phallynx of thumb.

2B- X-ray of chest did not show any hemivertebrae or rib anomalies .

Treatment- The goal is to achieve best possible functional use of hand[4].If associated with any cardiac and gastrointestinal anomalies , they require urgent surgical management and are given preference.For isolated cases management is conservative as well as surgical.

Parents are encouraged to passively stretch the wrist and hand to elongate the contracted radial soft tissues[4]. Serial casting and forearm splinting are applied at 1-2 weeks interval for 6 months[6].Surgery for correction of the wrist deformity remains controversial[4]. The surgery typically occurs when the child is 1 yr of age[6]. Correction of radial deviation as well as centralization of the wrist can be done by various surgical techniques which include open release, capsular reefing, tendon balancing[4].Pollicization for reconstruction of thumb with radial club[6]. The present case highlights about isolated longitudinal radial deficiency.

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