ISSN: 2455 – 9938
PubMed- National Library of Medicine - ID: 101731611

EIJO: Journal of Science, Technology and Innovative Research (EIJO–JSTIR)
Einstein International Journal Organization (EIJO)
Available Online at: www.eijo.in
Volume – 6, Issue – 6, November – December - 2021, Page No. : 23 - 27

Radial ray syndrome

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Type of Publication: Case Report

Conflicts of Interest: Nil

Abstract

Introduction: Radial ray syndrome is a rare congenital defect that may be isolated or associated with other anomalies. **Radial ray anomalies** comprise of a large spectrum of upper limb anomalies which range from partial (radial hypoplasia) to a complete (radial aplasia) deficiency of the radius with or without accompanying deficiency of the thumb bones.

Case Report: A 31 year old G3P1L1A1 with19+3 weeks of gestation presented to our hospital Chalmeda Anand Rao Institute Of Medical Sciences on 16/12/2021 with TIFFA scan report showing radial ray syndrome and termination of pregnancy was done in this patient

Conclusion: Radial ray malformation is an isolated finding which and can be unilateral or bilateral. Careful examination of the fetus will help to determine whether are any associated anomalies.

Keywords: Radial Ray Syndrome, Hypoplasia, Aplasia

Introduction

Radial ray anomalies comprise of a large spectrum of upper limb anomalies which range from partial (radial hypoplasia) to a complete (radial aplasia) deficiency of the radius with or without accompanying deficiency of the thumb bones. Radial ray anomalies and associated malformations range from unilateral, sporadic defects to bilateral defects that are associated with multiple malformation syndromes1.This rare malformation occurs in 1 in 30,000 live births[2].

Radial ray defect is a rare congenital that may be isolated or associated with other anomalies. Some of the well-known combinatonsare Fanconi pancytopenia syndrome, Holt-Oram syndrome, Thrombocytopenia-absent radius (TAR) syndrome, VACTERL syndrome. Detailed ultrasonography, including 2-D or3-D imaging, offers an opportunity to better appreciate subtle anomalies[3,4,5].

Case Report

A 31 year old G3P1L1A1 with19+3 weeks of gestation presented to our hospital Chalmeda Anand Rao Institute of Medical Sciences on 16/12/2021 with TIFFA scan report showing radial ray syndrome

Ductus venosus Doppler showed reversal of A wave in early weeks, on Tab. Ecospirin later it was normalized, no H/O of any teratogenic drug intake

TIFFA scan showed bilateral club foot with hypoplasia of bones of bilateral forearm with abnormal attachment of hand, fingers to hypoplastic forearms segment and abnormal four chambered heart with hypoplasia of left side of heart- likely, Radial Ray Syndrome

NIPT report showed high risk for TRISOMY 18

She is a k/c/o hypothyrodism, she is on Tab. Thyronorm 100mcg/OD since 7 years

In her first pregnancy, all trimester were uneventful, TIFFA was normal, delivered a live female baby by full term LSCS, baby is now 6 year old and healthy. She had induced abortion I/v/o absent fetal cardiac activity in her second pregnancy.

On examination

She is afebrile, her pulse rate was 80bpm, BP measuring 110/70mmHg, bilateral air entry was present and equal on both sounds, both heart sounds are heard.

On Perabdominal examination: Fundal height was corresponding to 18-20 weeks of gestation, fetal parts are palpable, fetal heart rate was 156bpm

Patient got admitted at our hospital Chalmeda Institute of Medical Sciences for further management

Her Hb was 11.4g/dL, platelet count was 3.11L/cumm, her CUE, RFT, LFT, HbA1C, PT, aPTT, INR was normal.

Termination of pregnancy was done.

X-ray of fetus was taken showed bilateral club hands and club foot, absence of thumb in left hand, hypoplastic radius in right forearm, complete absence of radius in left forearm.

Patient got discharged in a haemodynamically stable condition.

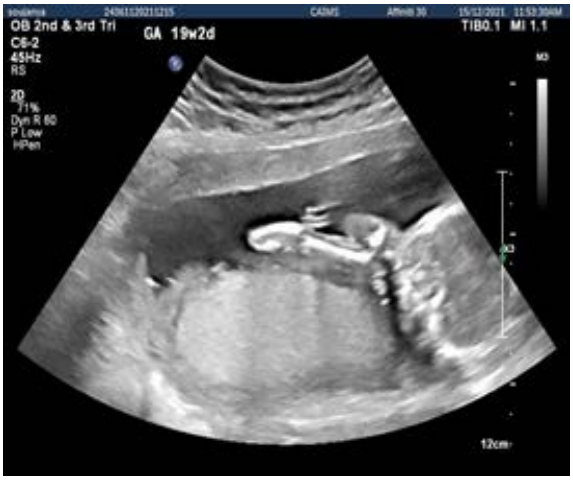

Figure 1: Radial deviation of right hand

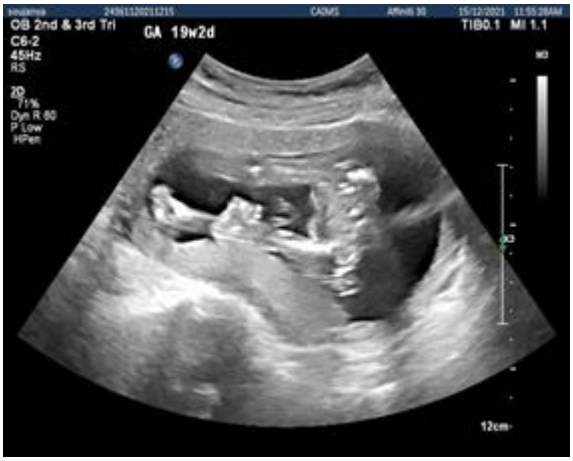

Figure 2: Hypoplastic radial bone in right arm

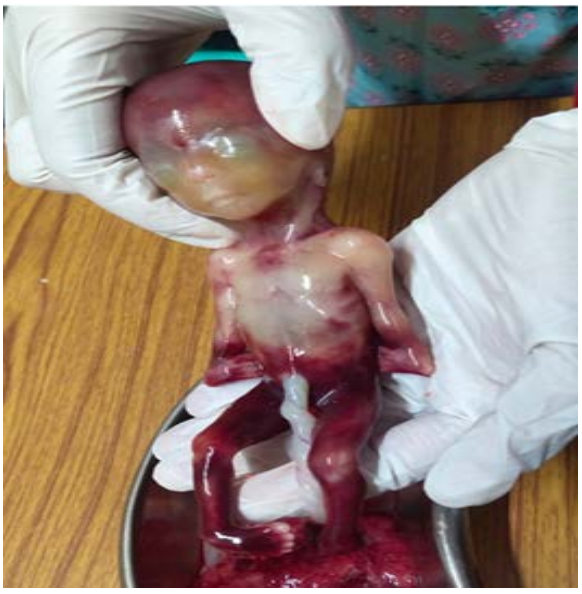

Figure 3: necropsy image bilateral club hands and club foots

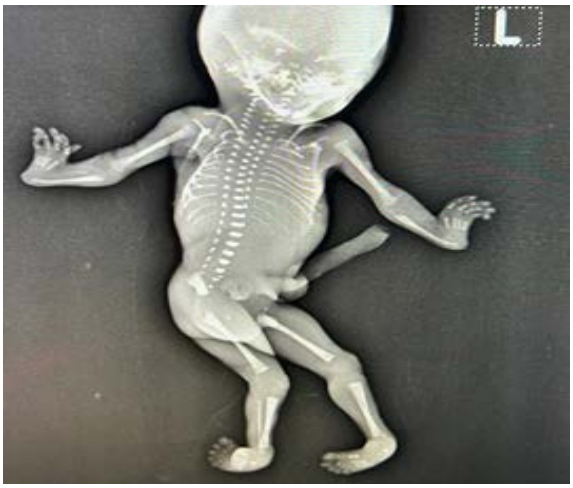

Figure 4: Postmortem x-ray

Discussion

The radius develops similarly to other appendicular bones through the process of endochondral ossification and develops from lateral plate mesoderm. Development of the limbs is under exquisite genetic control, and the genes that regulate the process are highly conserved through evolution. 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 underlying mesenchymal tissue and limb formation. Removal of a portion of the AER in chick embryos has produced anomalies similar to radial clubhand[6].

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[7]. Radial ray anomalies can be classified into four main subtypes depending upon the extent of severity:

- Type I: 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: 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

Some reports indicate that isolated RRD occurs in 8%-30% of all cases8.The majority of associated major congenital syndromes include VACTERL associations. The phenotype is largely variable, including certain anomalies of the vertebrae and limbs. Many cases are sporadic. A strong association exists between the heart and RRD, and is evident at birth. Holt-Oram syndrome (heart anomalies, and especially preaxial limb reduction), Fanconi pancytopenia syndrome (radial hypoplasia, hyperpigmentation, and pancytopenia),Roberts syndrome (upper limb reduction anomalies and heart de-fects), Bollere Gerold syndrome (craniosynosis and radial aplasia) and Cornelia de Lange syndrome (minor limb abnormalities, heartdefects, and hearing and eye problems) may be related to RRD[3,4,5,9-12].

When a radial ray anomaly is detected, these syndromes should be considered. Some reports have also proposed that valproate or thalidomide exposure might be associated with RRD[3,4,10].

Autosomal dominant, autosomal recessive, and X-linked inheritance have been proposed9. A detailed family history or exposure to a teratogen or drug should be included to obtain detailed consanguinity information. Fetal screening and prenatal diagnosis, including ultrasound and maternal serum screening or a genetic approach in early pregnancy, can detect trisomy 18, 13, 22q 11.2, and other syndromes.

The use of 3D ultrasound actually can facilitate the diagnosis of RRD, even at early gestation, because it provides a better surface appearance, panoramic view, and spatial orientation, which can be obtained to check the posture and configuration of the upper limbs. The deformity of the hands with radial angulation and loss or dysplasia of the thumb can be well-depicted on X-ray or necropsy findings. Genetic evaluation is by using Diagnostic tests (amniocentesis or chorionic villus sampling) with chromosomal microarray analysis (CMA) should be offered when a radial ray malformation is detected. Many syndromes are associated with radial ray malformations; they can be sporadic, autosomal dominant, autosomal recessive, or X-linked.

Management includes detailed ultrasound examination should be performed and should include assessment of all of the long bones and the hands. The fetal heart should be evaluated carefully, and a fetal echocardiogram should be considered

given the association with Syndromic disorders. Referrals to pediatric orthopedics or other subspecialty services should be based on additional sonographic findings. Pregnancy termination is an option that should be discussed with all patients in whom a fetal anomaly is detected. Shared patient decision-making requires a thorough evaluation and multidisciplinary counseling including orthopedic, plastic surgeon, psychologist taken regarding prognosis. The prognosis in isolated cases includes functional limitations that result from the skeletal deformity. Reconstructive surgery may improve the mobility of the affected limb.

Conclusion

Radial ray malformations are most commonly an isolated finding and can be unilateral or bilateral. Careful examination of the entire fetus will help determine whether there are associated anomalies. A genetic evaluation is recommended to determine the presence of an underlying syndrome. Prognosis depends on the severity of associated abnormalities and the presence of a genetic abnormality. Reconstructive surgery may improve limb mobility.

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