ISSN PRINT 2319 1775 Online 2320 7876

 UGC CARE Listed (Group -I) Journal Volume 11, S Iss 3,Dec 2022

NEWBORN WITH RADIAL DYSPLASIA AND SPINE DEFORMITY- AN INCOMPLETE VACTERL SYNDROME -A Rare Presentation

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Key word- Radial dysplasia, vertebral kyphoscoliosis, trachea-esophageal atresia, ASD

ABSTRACT

Radial dysplasia is a longitudinal deficiency of radial bone with abnormal bowing of fore arm. Usually, it is associated with syndromes and it is imperative to look out for other congenital anamolies[1]. We report a rare case of newborn born in our hospital with features like lumbar vertebral anomaly, cardiac defect (ASD) and limb anomalies like unilateral absent radius and absent thumb, bilateral CTEV. AS the baby had asymptomatic thrombocytopenia only on day 1 of life, by excluding the other congenital conditions like TAR syndrome, Fanconi anaemia, Holt Oram Syndrome etc, the baby is diagnosed to have Incomplete Vacterl syndrome with absence of trachea oesophageal fistula and anal atresia.

Keywords: Anal atresia, Radial dysplasia, Spine deformity, Vacteral syndrome

INTRODUCTION

Radial dysplasia, the term itself defines there is an abnormality in the radial bone. Radial dysplasia or radial club hand is a congenital malformation of radial fore arm which varies from radial hypoplasia to absent of radius. It is an uncommon condition in new born with predominance in male. Usually it is associated with other congenital conditions in the baby which needs to be evaluated to make a diagnosis of the child by ruling out other syndromic features. VACTERL syndrome is a group of congenital malformations with each word specifies unique congenital malformation; V-vertebral anamolies, A-anal atresia, C-cardio vascular anamolies, T- trachea oesophageal fistula, E – esophageal atresia, R- renal /and radial anamolies, L- limb defects. Having a minimum of three findings is required to suggest the child is having VACTERL syndrome and can be confirmed only by excluding other syndromes with similar clinical or morphological features.

CASE REPORT

A male baby born to a via normal spontaneous vaginal delivery, to a non consanginous married couple. Cried soon after birth with thin MSL all over the body. Baby cried soon after birth with no birth asphyxia. Baby had mild tachyapnea. Injection vit K 1.00 mg given IM immediately after birth.

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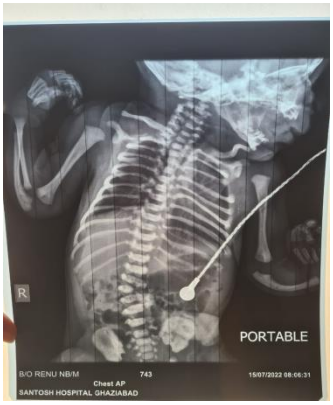
On examination, baby had club forearm and hand with absent of thumb finger and fifth metacarpal bone (Legend 1). No anorectal or spinal deformity visible at the time of birth.

Baby shifted to NICU for observation &kept on oxygen support via hood. Baby had mild respiratory distress which is settled in 24hrs.After 36 hrs baby vitals were stable and accepting breast feed well and passing urine and stool normally.

Anthropometry: weight -2.9kg, length - 50 cm, Head circumference- 35 cm

Xray of Chest, Abdomen and B/L upper limbs are taken

Legend 1-showing left fore arm with club hand



Legend 2 – showing kyphoscoliosis with hemi lumbar vertebral anamoly

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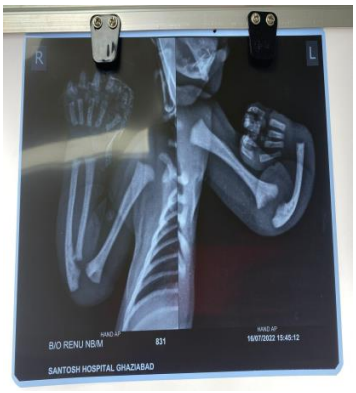

Legend -3 X- ray of left forearm showing absent of radius ,fifth metacarpal and thumb

To exclude other anomalies, the skeletal survey of was done and and consistent with TYPE IV(as described by Heikel) Radial Dysplasia (legend-1) (complete absent of radius, metacarpal bone and thumb, with severe bowing of ulna wrist and hand towards radial side (legend- 3)

Chest and abdominal x-ray shown anomaly in lumbar vertebrae (legend- 2)

Baby had B/L CTEV of lower limbs.2D ECHOCARDIOGRAPHY – showed ASD

Other Investigations showed- Hb – 20.5gm%, platelets – 65,000/cu mm, TLC- 23420/cu mm, DLC- Neutrophils-73%, Lymphocytes-22%, Monocytes-5%, CRP -1.52 mg/L, Platelet count improved on day 5 of life and rose to 3.30 Lac/mm^3

Both mother and baby were A+ve,LFT & KFT are with in normal levels , NCCT of head was normal.

DISCUSSION

VACTERL is a heterogenous condition with multiple birth defects like vertebral anamoly, anal atresia, cardiac anomaly, trachea esophageal fistula , esophageal atresia, limb and renal anamolies. Whereas our baby had 3 anomalies namely, 0

left radial aplasia, vertebral anomaly and heart defect[2].VACTERL is kept as diagnosis by exclusion i,e ruling out other syndromes who have radial dysplasia like HOLT ORAM syndrome which has bilateral lupper limb anomalies[3] Fanconi anaemia is ruled out as the child had no microophthalmia, strabismus ,or hearing defects[4]. Cornelia de Lange syndromeis ruled out as the baby had no micromelia and absence of hairs all over body.TAR syndrome, is ruled out as the platelets increased by day 3, unilateral absent of radii and baby had absent thumb[5].Our patient doesn't have any renal anamoly or esophageal fistula baby was given supportive treatment and by day 5 started accepting feeds and discharged with an advice to take an opinion from orthopaedic surgeon regarding limb reconstruction and progressives in growth and functional.

Family history was normal

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Limitation of study - Genetic workup was not done.

Abbreviations used in study

VACTERL, Holt Oram, Fanconi,TAR are name of syndromes.

CTEV-Congenital TalipesEquinoVarus, LFT-liver function test, KFT-kidney function test

Conflict of Interest-None

Funding- None

CONTRIBUTION BY AUTHORS

DR.Naveen Nelakurthi : Data acquisition, analysis ,work up of case

DR.K.C Aggarwal: Guide and editor of the literature,

DR.Mannat Verma :Reviewed the articles

DR.Jyoti Batra: Drafting of the manuscript

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