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Original Research Paper

Paediatrics

RADIAL DYSPLASIA AND SPINE DEFORMITY OF NEW BORN – INCOMPLETE VACTERL SYNDROME

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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 anamolies1 . We report a rare case of newborn born in the hospital with features like lumbar vertebral anomaly, ASD and limb anomalies that is unilateral absent thumb and absent radius, bilateral Congenital Talipes Equino Varus. AS the baby had asymptomatic thrombocytopenia only on day 24hrs of life, by excluding the other congenital conditions like Thrombocytopenia-absent radius 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 : Vacterl syndrome, Spine deformity, Radial dysplasia, Anal atresia

INTRODUCTION

Radial dysplasia, the term itself defines there is an abnormality in the radial bone. It is an uncommon condition in new born with predominance in male. Radial dysplasia or radial club hand is a congenital malformation of radial fore arm which varies from radial hypoplasia to absent of radius. 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 & 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 Study

A male baby born to a via normal vaginal delivery, to a third degree consanginous married couple. Baby cried immediately after birth with thin MSL over the body. Baby cried soon after birth with no birth asphyxia. Baby had moderate tachyapnea. Injection vit K 1.00 mg given Intramuscular immediately after birth.On examination, baby had hand with absent of thumb finger, club forearm and fifth metacarpal bone (Fig 1). No anorectal or spinal deformity visible at the time of birth. Baby shifted to NICU for observation & kept on oxygen support via high flow nasal cannula. Baby had moderate respiratory distress which is settled in 36hrs. After 48 hrs baby vitals were stable and accepting breast feed well and passing urine and stool normally.

Anthropometry: weight -2.6kg, length - 48 cm, Head circumference- 34 cm X ray of Chest, Abdomen and Bilateral upper limbs are taken

FIG 1-showing left fore arm with club hand

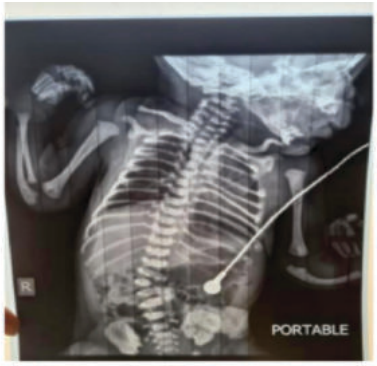

FIG 2 – showing kyphoscoliosis with hemi lumbar vertebral anamoly

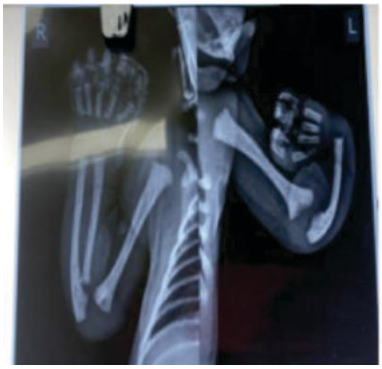

FIG -3 X- ray of left forearm showing fifth metacarpal, absent of radius and thumb

To exclude other anomalies, the skeletal survey of was done and consistent with TYPE IV (as described by Heikel) Radial Dysplasia (Fig -1) complete absent of radius, metacarpal bone and thumb, with severe bowing of ulna wrist and hand towards radial side (Fig - 3) Chest and abdominal x-ray shown anomaly in lumbar vertebrae (Fig - 2) Baby had Bilateral Congenital Talipes Equino Varus of lower limbs.

2D ECHOCARDIOGRAPHY – showed small ASD

Other Investigations showed- Hb – 18.5gm%, platelets – 60,000/cu mm, TLC- 20480/cu mm, DLC- Neutrophils-70%, Lymphocytes-25%, Monocytes-5%, CRP -2.65 mg/L, Platelet count improved on day 7 of life and rise to a value of 4.30

Lac/mm^3 both mother and baby were B+ve, LFT & RFT are with in normal levels , Non contrast computed tomography of head was normal.

DISCUSSION

VACTERL is a heterogenous condition with multiple birth defects like vertebral anamoly, trachea esophageal fistula, anal atresia, cardiac anomaly, esophageal atresia, limb and renal anamolies. Whereas our baby had three anomalies that are, vertebral anomaly, left radial aplasia, 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 micro ophthalmia, strabismus ,or hearing defects[4] . Cornelia de Lange syndromeis ruled out as the baby had no micromelia and absence of hairs all over body. Thrombocytopenia-absent radius 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 4 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

Limitation of study – Genetic studies not done.

Abbreviations used in study VACTERL, Holt Oram, Fanconi, Thrombocytopenia-absent radius are name of syndromes. Congenital Talipes Equino Varus, liver function test, kidney function test

Conflict of Interest-None

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