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Case Report

Neonate with VACTERL association: a rare entity

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ABSTRACT

VACTERL (Vertebral, anal, cardiac, tracheoesophageal, renal and limb) is an acronym for cluster of congenital malformations including Vertebral or Vascular anomalies, Anal atresia, Cardiac defects, Tracheoesophageal fistula/Esophageal atresia, Renal and Limb defects. It is a rare association with sporadic and non-random occurrence where multiple organs are affected due to developmental defect during blastogenesis. Exact cause is unknown but multiple environmental and genetic factors have been implicated. For diagnosis, three components of VACTERL association are needed however patient may have other congenital malformations also. Here we report a newborn with VACTERL association born to a healthy mother by cesarean section for polyhydramnios with unstable lie.

Keywords: Limb defects, Polyhydramnios, Tracheoesophageal

INTRODUCTION

VACTERL association is a constellation of vertebral (V), anal (A), cardiac (C), Tracheoesophageal (TE), renal (R) and limb (L) anomalies in a single individual. Heterogeneous clinical profile with no definite gene defect defines this condition as an association rather than a syndrome.[1] VACTERL association is a complex and rare entity with reported incidence between 1/10000 - 1/40000.[2] It is diagnosed by the simultaneous presence of at least three out of the six VACTERL acronym anomalies without clinical or laboratory evidence suggesting other differential diagnosis.[3]

CASE REPORT

A baby girl was born with anomalous right upper limb (absent thumb and shortened forearm, Figure 1) at 40 weeks of gestation by emergency cesarean section. Mother was 26-year-old second gravida with non-consanguineous marriage to a 28-year-old man. There was no family history of birth defects and first sibling was completely normal.

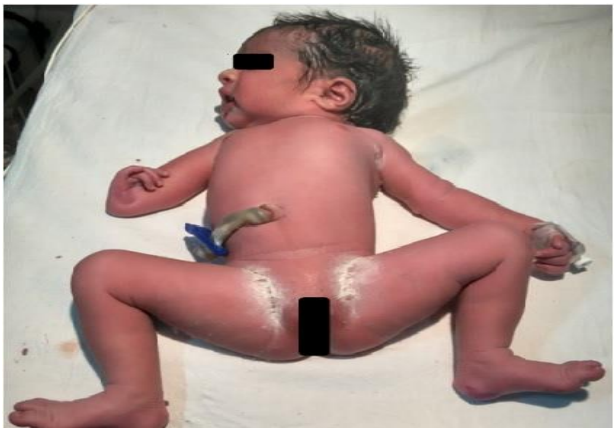

Figure 1: Female neonate with absent right thumb and shortened right forearm.

But mother revealed a significant history of hyperemesis gravidarum and recurrent urinary tract infections in the first trimester of this pregnancy. She had taken multiple treatments from govt. and private sectors but records couldn't be traced out to establish a particular drug or intrauterine infection as a cause for VACTERL association. Further mother didn't give any history of smoking, alcohol intake, radiation exposure and diabetes during antenatal period. She was a booked case but unfortunately anomaly scan was missed at 20th weeks of gestation. Emergency cesarean section was conducted in view of fetal distress and polyhydramnios with unstable lie. Baby cried immediately after birth with Apgar score of 8/10 at one minute and 9/10 after five minutes. All orifices (oral, nasal, vaginal and anal) were patent and no other gross anomalies were found on physical examination. The anthropometric measures of baby revealed birth weight of 2.6kg, length 46cm and head circumference 34cm. Approximately 20 minutes after birth, profuse frothing was noticed from the mouth of baby. So, a nasogastric tube was inserted but it couldn't pass down. Simultaneously an infantogram, USG abdomen, ECHO studies and routine blood investigations were advised to screen out other systemic anomalies. Infantogram showed absent radial ray bones (radius, metacarpal and phalangeal bones) of right side along with oesophageal atresia as evidenced by coiling of nasogastric tube (Figure 2).

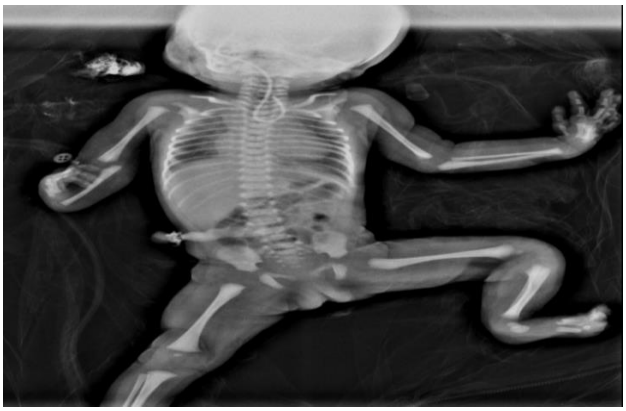

Figure 2: Infantogram showing Esophageal atresia as evidenced by coiling of nasogastric tube (yellow arrow) and absent right radial ray bones (red arrow).

Vertebral and rib bones were normal on scanning. USG abdomen revealed absence of left kidney while on ECHO studies, small ventricular septal defect (VSD) was noticed. Rest of the systemic evaluation was within the normal limits. Further routine blood tests (complete blood counts, peripheral blood smear, renal function tests, electrolytes, serum calcium and blood sugar) were also within the normal range. So, based on absent radial ray bones, esophageal atresia, VSD and absent left kidney, diagnosis of VACTERL association was established. Further normal blood investigations and systemic evaluation ruled out the other differential diagnosis of VACTERL association. After stabilization, baby was referred to higher centre for further management under the cover of oxygen and intravenous fluids.

DISCUSSION

In 1973, Quan and Smith were the first to describe the VATER association.[4] Temtamy et al suggested the inclusion of vascular defects like ventricular septal defect (VSD) and single umbilical artery in the V of VATER association.[4] In 1974, addition of cardiac (C) and limb defects (L) changed the acronym to VACTERL association. Recently VACTERL-H has been described for associated hydrocephalus (H) in the newborn.[5]

The etiology of VACTERL association is unknown and many cases occur sporadically. Several authors have suggested that developmental field defect during blastogenesis (2-4 weeks of gestation) may impair the formation of multiple organs simultaneously.[6] Multiple environmental factors like maternal diabetes mellitus, alcohol and teratogenic drugs (sex hormones, anticholestrol drugs, lead, adriamycin, venlafaxine, anti-epileptic drugs) interact with genetically susceptible genome.[6] Although the exact genetic cause has not been established but several genetic mutations, chromosomal aberrations and mitochondrial defects has been reported in relation to some components of VECTERL association.[5] Most of the causes were ruled out in our case but we couldn't establish the exact cause in spite of having exposure to multiple drugs and infective pathogens in the first trimester of pregnancy.

Vertebral, anal, cardiac, trcheoesophageal, renal and limb anomalies constitute the major components of VACTERL association. In addition to these prime features, patients may also have other congenital anomalies like hydrocephalus, ribs and brachial arch anomalies. Our baby had the four (cardiac, tracheoesophageal, renal and limb) major components of VACTERL association. Vertebral anomalies are the most frequently reported defects in 60-80% cases but they were not appreciated in our case.[7] Renal anomalies are reported in 50 to 80% of the cases in the form of unilateral renal agenesis (absent left kidney in our case), horseshoe, and cystic and/or dysplastic kidneys.[8] Cardiac anomalies occur in 50-80% of cases further VSD, ASD, (finding of our case) and tetrology of Fallot are amongst the frequently reported defects.[3] Tracheo-esophageal anomalies occur in approximately 50-70% of patients and they may be associated with lung anomalies. Limb anomalies are reported in approximately 40-55% patients and include radial ray deformities, (absent radius, polydactyly, syndactyly, radioulnar synostosis) club foot, hypoplasia of great toe/tibia and lower limb tibial deformities.[8]

As VACTERL association is diagnosis of exclusion, so we excluded rare syndromes like Alagille syndrome (no typical facial appearance or ophthalmic anomalies), Baller-Gerold syndrome (no skin anomalies or

craniosynostosis), CHARGE syndrome (no coloboma, ear anomalies /facial features), Fanconi's anemia (no abnormality in pigmentation or hematologic parameters), Feingold's syndrome (no syndactyly at toes) and Fryns' syndrome (no diaphragmatic abnormality or facial anomalies) on clinical evaluation and investigative workup.[6]

Some components of VACTERL association can be diagnosed quite early by antenatal imaging, prenatal echocardiography and MRI, but unfortunately anorectal and tracheo-esophageal anomalies often may not be detected in the second and early third trimester of pregnancy, even with skillful examinations.[7]

The treatment of VACTERL association includes surgical correction of severe cardiac defects, imperforate anus, tracheaesophageal fistula and limb defects. Further long-term medical management of the renal and vertebral co-morbidities along with rehabilitation is required. Overall prognosis is not good so if detected in- utero, termination of pregnancy is advisable. Further, in postnatal period early detection and early surgical intervention along with lifelong rehabilitation can improve the outcome.[8]

CONCLUSION

VACTERL association is a complex and rare entity with random occurrence of multisystem congenital malformations in a single patient.

In those setups (like ours) where genetic studies are not available a detailed antenatal history regarding triggering factors can give a clue regarding etiology. Skillful antenatal screening along with careful postnatal clinical as well as investigative evaluation for systemic anomalies in an anomalous newborn is required to diagnose rare entities like VACTERL association.

Early detection and timely referral for surgical interventions can improve the prognosis of infant with VACTERL association.

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