MEDICAL SCIENCE

A rare case of VACTERL association in new born

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ABSTRACT

VACTERL Association is complex, non-random sporadic association of various birth defects. It includes vertebral, anal anomalies, cardiovascular, tracheoesophageal, renal anomalies, radial anomalies and limb abnormalities. It is first described in 1970s. Full spectrum association is a very rare entity. VACTERL is considered to be present if any three of the above-mentioned abnormalities are present. Along with these features, patients might present with other external anomalies like lung lobation defects, external ear anomalies etc. However, the full spectrum of VACTERL association is very rare diagnosis mainly by clinical approach. Multidisciplinary approach and treatment of surgically correctable anomalies provide a better outcome for this association in new born period. Tracheoesophageal fistula is associated in more than 70 percent of the cases. Limb defects are also present in 70 percent of the cases. Anal atresia is present in 55 percent of the cases. VACTERL Shows phenotypic overlap with various conditions like Goldenhar syndrome, Fanconi anaemia, CHARGE syndrome etc. Here in we are reporting a case of VACTERL with presence of cardiovascular abnormality, Tracheoesophageal fistula and limb abnormalities.

Keywords: Cardiovascular abnormality, Tracheoesophageal fistula, lung lobation

1. INTRODUCTION

Globally congenital anomalies account for about six percent and 94% amongst them are present in the developing countries. The most common birth defect among all is congenital heart diseases. VACTERL association is first discovered in early 1970. It has no random and sporadic association. It includes V-Vertebral, A-Anorectal malformation, C-Cardiac, T-Tracheoesophageal, R-Radial/Renal abnormalities, L-Limb abnormalities. Incidence of this association is very rare that is 1 in 10000 (Khoury et al., 1983). Presence of at least three of the abovementioned features is considered to be as positive association however full spectrum of VACTERL is a very rare entity. It is associated with other abnormalities like dysmorphic features, lung lobation defects, external ear abnormalities etc. (Solomon, 2011). Here in we are reporting a neonate with cardiac abnormality (ASD with bi directional shunt), tracheoesophageal fistula, radial abnormality and limb anomaly.

2. CASE REPORT

A 1.7 kg male baby born to a G2P1L1 mother with 38 weeks of gestational age. It was a twin gestation. The baby was delivered via lower segment caesarean section, given leaking per vagina and no progress of labor. The baby cried immediately after birth and on passing the nasogastric tube during the examination of the neonate, there was an inability to pass the nasogastric tube. X-ray done with the nasogastric tube in situ was suggestive of coiling of the nasogastric tube, which indicates the presence of a tracheoesophageal fistula (Figure 1). The patient was referred to our center given.

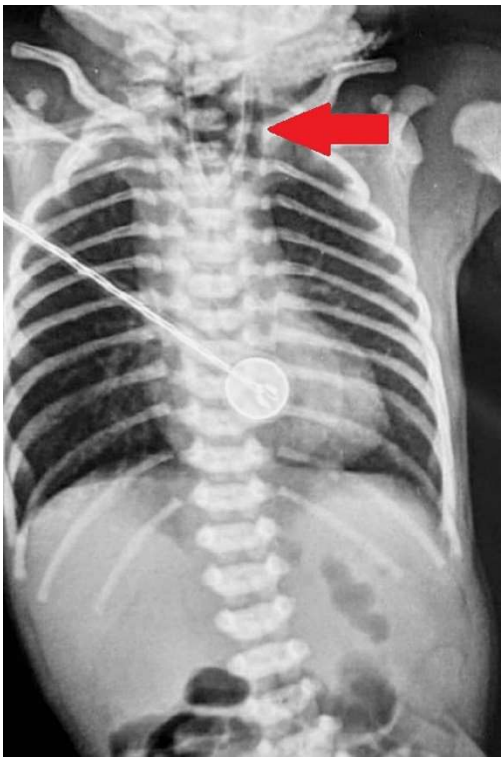

Figure 1 X-ray showing coiling of nasogastric tube suggestive of tracheoesophageal fistula

On examination, cry and tone activity was good, HR 140/min, RR 38/min, peripheral pulses were well felt and Spo2 was 98 percent in room air. On examination forearm appears to be reduced or absent, the hand is supinated or abducted over the arm and a thumb is absent in hand (Figure 2). On auscultation, air entry was bilaterally equal; there was a soft mid-systolic murmur and an inability to pass the nasogastric tube. Per abdomen was soft and the liver and spleen were not palpable.

Basic investigations were sent s/o HB 19.9, Total Leukocyte Count 11200, Platelet 2.8 lakhs, C-reactive protein 1.2 and liver function test and kidney function test were in the normal range. The patient was put on oxygen with nasal prongs and injection cefotaxime and injection amikacin were started. Chest x-ray was done s/o coiling of the nasogastric tube, which was reported as tracheoesophageal fistula. X-ray of the right hand was done s/o absent radius (Figure 3); Echocardiography was done s/o ostium secundum-atrial septal defect with bi-directional shunt, mild dilated right atrium and right ventricle. Neuro sonogram was normal. There were no vertebral anomalies, anorectal abnormalities and renal abnormalities.

On the day of life two babies was operated for right posterolateral thoracotomy with Ligation of tracheoesophageal fistula type C with esophagostomy, gastrostomy (Figure 4) and inter-costal drain insertion (Figure 5). Intra-op was uneventful. Post op patient was shifted to NICU intubated. Antibiotics were upgraded to meropenem, colistin and fluconazole. The patient was extubated on the day of surgery two and was taken on CPAP. Intralipid infusions were started, gastrostomy feeds were started post-extubation, esophagostomy tube and Inter Coastal Drain were slowly removed. The orthopedic call was made and the mother was advised to do passive stretching exercises towards the ulnar side and review after one month. A cardiology opinion was taken and was recommended for device closure after three months. Full gastrostomy feeds were established and the baby was discharged.

Figure 2 Forearm appears reduced or absent, hand is supinated and abducted over the arm and absence of thumb in right hand

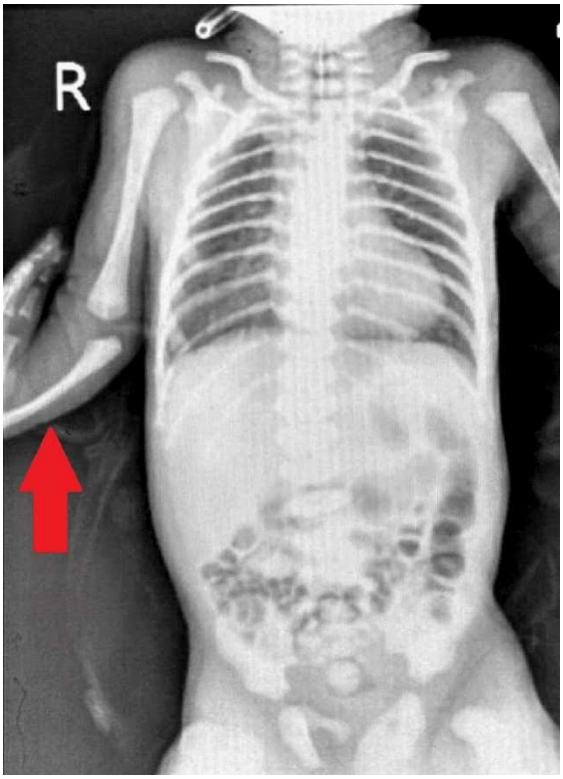

Figure 3 X-ray showing absent radius

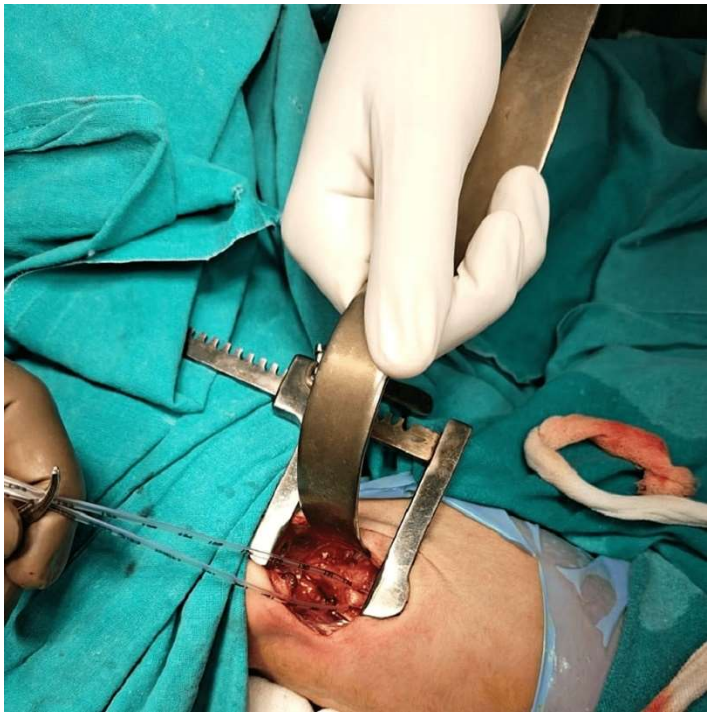

Figure 4 Intraoperative image of tracheoesophageal fistula repair

Figure 5 Esophagostomy tube insertion intraoperative tracheoesophageal fistulas

3. DISCUSSION

The incidence of VACTERL association is 1 in 10000 to 1 in 40000. It is a non-random sporadic association of various congenital disabilities of structures arising from embryonic mesoderm. VACTERL, with all the associated anomalies described, is very rare and is present only in 1 percent of the cases (Botto et al., 1997). It was first described in 1972 (Mc-Cartney and Marshall, 2016). Among the further anomalies described, at least three should be present to label it as a VACTERL association (Harjai et al., 2008). The mechanism of VACTERL is not completely understood. There might be a recurrence of this association in subsequent pregnancies also. Careful antenatal screening should be performed. Early detection of the anomalies and a multidisciplinary approach with proper timing of surgeries like anal atresia, tracheoesophageal fistula and cardiovascular abnormalities and correction of a few corrective orthopedic abnormalities are associated with good outcomes.

About 70 percent of VACTERL association V-Vertebral anomalies consist of vertebral anomalies, including hemivertebrae or small vertebrae. In the child's later life, there might be a risk of acquisition of scoliosis. A-Anal abnormalities it is present in anal atresia and imperforate anus. C-Cardiovascular abnormalities may be associated with cardiovascular abnormalities like Atrial Septal Defect, Ventricular Septal Defect and in rare cases, Tetralogy of Fallot. T-Tracheoesophageal fistula about 70 percent of the cases is associated with oesophageal atresia with trachea oesophageal fistula. R-Renal anomalies are associated with unilateral or bilateral renal abnormalities with obstructive uropathy. About 70 percent of L-Limb defects are associated with forearm abnormalities, absent thumb, polydactyly or syndactyly and radial aplasia.

It can prenatally be detected in a few cases by the absence of stomach bubbles, dilated colons and limb anomalies. There is no specific diagnostic test for the diagnosis of VACTERL. It is a clinical diagnosis based on anomalies (Salati and Rabah, 2010). Management of the associated surgical complications like anal atresia and tracheoesophageal fistula provides a good outcome. Management is by multidisciplinary approach by pediatrician, pediatric surgeon, orthopedics, otorhinolaryngology specialist, Physiotherapist and Urologist (Arsic et al., 2002; Aynaci et al., 1996; Quan and Smith, 1973). VACTERL has many overlapping phenotypical features with other syndromes like Fanconi anemia, Goldenhar syndrome, CHARGE syndrome, etc. (Källén et al., 2001). In previous study described VACTERL association with the associated cardiac anomaly, tracheoesophageal fistula and anal atresia. Chauhan et al., (2017) described the VACTERL association with tracheoesophageal fistula, radial and cardiac abnormalities. In previous study described full spectrum VACTERL association with other associated dysmorphic features.

4. CONCLUSIONS

VACTERL association is a non-random and sporadic association of abnormalities in various systems. Careful antenatal evaluation is performed and if severe anomalies are detected, parents should be counselled for the termination of pregnancy. There might be a recurrence of this association in subsequent pregnancies also. Careful antenatal screening should be performed. Early detection of the anomalies and a multidisciplinary approach with proper timing of surgeries like anal atresia, tracheoesophageal fistula and cardiovascular abnormalities and correction of a few corrective orthopedic abnormalities are associated with a good outcome.

Informed consent

Written & Oral informed consent was obtained from participant parents included in the study.

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Conflict of interest

The authors declare that there is no conflict of interests.

Data and materials availability

All data sets collected during this study are available upon reasonable request from the corresponding author.

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