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Case Report

VACTERL Association: A Case Report in Newborn

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Abstract: VACTERL association is a useful acronym for a condition characterized by the sporadic, non-random association of specific birth defects of multiple organ systems. It includes vertebral anomalies (V), anal anomalies (A), cardiac anomalies(C), trachea-esophageal fistula (TE), R-renal anomalies or radial bone anomalies(R), L-limb defect (L). Association is defined by presence of at least three of above malformation. In addition to these core component features, patient may also have other congenital anomalies. We are reporting one such case which had congenital abnormalities of cardiac system, trachea-esophageal fistula and vertebral anomalies. Purpose of this case report is to highlight the distinctive presentation of VACTERL association, to enable earlier detection, to discuss the treatment option.
Keywords: VACTERL, anomalies, malformation

INTRODUCTION:

VACTERL association is a mnemonically useful acronym for a condition characterized by the sporadic, non-random association of specific birth defects of multiple organ systems. Described in the early 1970s, VACTERL association is typically defined by the presence of at least three of the following congenital malformations - vertebral defects, anal atresia, cardiac defects, and treacheo-esophageal fistula, renal anomalies and limb defects. In addition to these core component features, patients may also have other congenital anomalies like hemifacial microsomia, external ear malformations, lung lobation defects, intestinal malrotation and genital anomalies [1]. Incidence is estimated at approximately 1 in 10000 to 1 in 40000 live births [2].We present one such case which had congenital abnormalities of cardiac system, tracheo-esophageal fistula and vertebral anomalies.

CASE REPORT

A five hour preterm, first order male child born to nonconsanguineously married couples was brought in N.I.C.U. of our institute with complaint of poor feeding, excessive drooling of saliva and fast breathing. He was born at gestational age 37 wks by L.S.C.S following complaint of polyhydramnios in mother. During this pregnancy the mother was registered and immunized. She underwent regular antenatal check-ups, she underwent thrice obstetric scan in her pregnancy which was told to be normal and there was no significant past medical or surgical history. There was no family history of congenital anomalies. On examination baby was sick looking. He weighed 2.3kgs, had tachypnea with increased work of breathing and spO2 was maintained with 5 ltrs of oxygen. There was persistent frothing from the mouth and we were unable to insert a nasogastric tube into the stomach(Fig-2).

There was systolic murmur better heard on pulmonary area. On further evaluation and imaging abnormalities involving other systems were detected. Chest radiogram showed features of cardiomegaly, dextrocardia (cardiothoracic ratio greater than 50%), coiling of nasogastric tube at level of T4 with whole stomach filled with air shadow, hemivertebra at a level of T8 (fig-1) echocardiogram revealed moderate atrial septal defect with pulmonary valve stenosis (severe) and good systolic LV function. Ultra sonogram of abdomen found normal. Hemoglobin, total and differential leucocytes count, platelet count, and renal function tests were all within normal limits. Due to simultaneous occurrence of congenital anomalies involving cardiac, skeletal (vertebrae) and esophageal atresia with tracheoesophageal fistula the patient was labeled as a case of VACTERL association. The patient was managed in department of pediatric surgery.

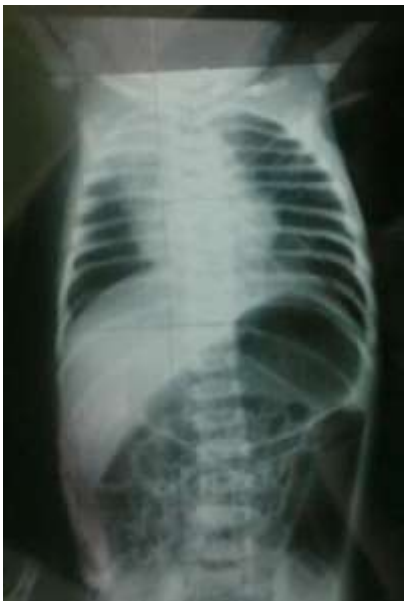

Fig-1: xray showing dextrocardia, abdominal distention, hemivertebra

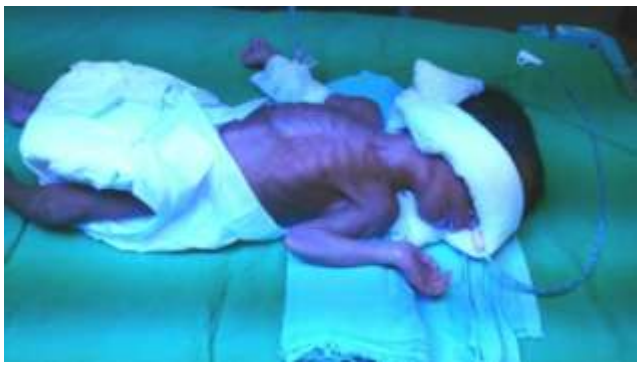

Fig-2: neonate with frothing from mouth, NG tube partially inserted

DISCUSSION

VACTERL association is a mnemonically useful acronym for a condition characterized by the sporadic, non-random association of specific birth defects in structures derived from the embryonic mesoderm. Each letter in VACTERL represents the first letter of one of the more common findings seen in affected cases. VACTERL association was first reported by Corcora et al. in 1975, [3] but only 1.0% of such cases present the full range of anomalies[4]. For getting labeled as VACTERL, there should be at least three out of the following seven findings[5].

V - Vertebral anomalies: Vertebral anomalies usually consist of hypoplastic(small) vertebrae or hemivertebra (where only one half of the bone is formed). About 70 percent of patients with VACTERL association will have vertebral anomalies. In early life these anomalies rarely cause any difficulties, although the presence of these defects on a chest x-ray may alert the physician to other defects associated with VACTERL. Later in life, these vertebral anomalies may put the child at risk for developing scoliosis.

A - Anal atresia: Anal atresia or imperforate anus is seen in about 55 percent of patients with VACTERL association.

C - Cardiovascular anomalies: Up to three quarters of patients with VACTERL association have been reported to have congenital heart disease. The most common heart defects seen with VACTERL association are ventricular septal defects, atrial septal defects and Tetralogy of Fallot. Less common defects are truncus arteriosus and transposition of the great arteries.

TE - Tracheoesophageal fistula: Esophageal atresia with tracheo-esophageal fistula (TE fistula) is seen in about 70 percent of patients with VACTERL association.

R - Renal (Kidney): Renal defects are seen in half the patients with malformation of one or both kidneys or obstructive uropathy.

L- Limb defects: Limb defects seen in up 70 percent of babies include absent or displaced thumbs, polydactyly, syndactyly and forearm (including radial aplasia) and leg defects.

In addition, to the above mentioned features, affected children may also exhibit less frequent abnormalities including growth deficiencies and failure to gain weight and grow at the expected rate (failure to thrive). Practically every organ system have been reported in association with VACTERL in lower frequency like facial asymmetry (hemifacial microsomia), external ear malformations, lung lobation defects, intestinal malrotation and genital anomalies[6].

VACTERL shows some pheonotypic overlap with many other conditions including Feingold syndrome, CHARGE syndrome, 22qll deletion syndrome, Townes-Brocks syndrome, Pallister-Hallsyndrome, Fanconi anemia spectrum, Goldenhar Syndrome, Nager syndrome, caudal regression syndrome, sirenomelia, electrodactyly-ectodermal dysplasia syndrome, Jarcho-Levin syndrome and Klippel- Fiel syndrome. Some researchers have added an (S) to the VACTERL acronym to represent a single umbilical artery instead of the normal two. Mental functioning and intelligence is usually unaffected[6].

The incidence of VACTERL association is not known exactly because of its wide range of manifestations; however, with the available literature the incidence is estimated to range from 1in 10000 to 1 in 40000 live births [2].

The etiology is currently unknown, but is believed to be multifactorial [8]. The combination of VACTERL abnormalities can present with some known chromosomal abnormalities, including trisomy 13, 18, and 5p- syndrome. Interstial deletion of long arm of

chromosome 6 (6q13-15) and long arm of chromosome 13 have been reported in few cases [9]. Recent research has shown that VACTERL could be caused by defective Shh (Sonic hedgehog pathway) signaling during human embryogenesis [8]. Though chromosomal abnormalities are reported in children with this association, it is rarely seen more than once in the family. The reason it is called an association rather than a syndrome is that while all of the birth defects are linked, it is definitely unknown which genes or sets of genes cause these birth defects to occur. A disruption in the differentiating mesoderm in the first 4-5 weeks after conception (during blastogenesis) has been suggested to be the basis for such a non-random association[4].

Diagnosis is mainly clinical and is based on the phenotypic features. Because the cause of VACTERL association is unknown, no laboratory test exists than can diagnose or rule out this condition[7]. As with many other conditions, the ability to detect features of VACTERL association prenatally, whether through ultrasound or more sophisticated methods such as prenatal echocardiogram or MRI is very much dependent on the skill and experience of the medical interpreter. Polyhydramnios, absence of a gastric bubble, dilated colon, vertebral defects and limb abnormalities are certain subtle radiological features that may suggest an affected fetus[8].

Treatment is directed towards the specific symptoms that are apparent in each child, which often varies greatly. Many of the structural abnormalities (radial defects, cardiac defects, anal atresia etc.) require staged surgical corrections. Infants with this condition need to be managed by a multidisciplinary team including pediatricians, cardiologists, urologists, orthopedic surgeons, otorhinolarngologists and clinical geneticist in order to have a reasonable life expectancy[9].

Prognosis for children with this condition depends on the severity of anomalies. With improvements in surgical techniques and in specialized neonatal and post-surgical facilities, these children have a much better outcome than reported previously. Nonetheless, even with optimal surgical management of cardiac defects, trachea-esophageal fistula, and limb abnormalities patients can face considerable medical challenges throughout life. Finally, despite significant morbidity associated with the component congenital malformations, it is also important to note that these patients do not typically display neurocognitive impairment[2, 10].

CONCLUSION

VACTERL association is a condition characterized by nonrandom association of specific birth defects involving multiple organ systems. Occurrence is usually sporadic. Etiology is multifactorial. Diagnosis is essentially clinical and requires defects in at least 3 organ systems as mentioned previously. Multidisciplinary management is required for these cases, with staged surgical therapy being the mainstay of treatment. Since the exact genetic basis for this condition has not yet been established, parents with an affected child must be reassured that the recurrence risk in subsequent pregnancies is extremely low and if detected early in utero before viability, termination can be offered.

REFERENCES:

1. Khoury MJ, Cordero JF, Greenberg F, James LM, Erickson JD; A population study of the VACTERL association: evidence for its etiologic heterogeneity. Pediatrics, 1983;71:815–20.
2. Benjamin DS; VACTERL /VATER Association. Orphanet Journal of Rare Diseases, 2011;6: 56.
3. Corcoran R, Entwistle GD; Letter: polycystic ovarian disease and oral contraception. Lancet, 1975; 21:1386–87.
4. Botto LD, Khoury MJ, Mastroiacovo P , Castilla EE, Moore CA, Skjaerven R et al.; The spectrum of congenital anomalies of the VATER association: An international study. Am J Med Genet, 1997;71:0815-20
5. Harjai MM, Holla RG, Kale CR; Full spectrum of VACTERL in new born MJAFI, 2008; 64:84-85
6. Kallen K, Mastroiacovo P, Castilla EE, Robert E, Kallen B; VATER non-random association of congenital malformations: Study based on data from four malformation registers. Am J Med Genet, 2001;101:26-32
7. Salati SA, Rabah SM; VACTERL association. Online J Health Allied Scs, 2010; 9(2): 15.
8. Arsic D, Qi BQ, Beasley SW; Hedgehog in the human: a possible explanation for VATER association. J the Paediatr Child Health, 2002; 38(2): 117-21.
9. Aynaci FM, Celep F, Karaguzel A, Baki A, Yildiran A; A case of VATER association associated with 9qh+.Genet Couns, 1996; 7(4): 321-322.
10. Quan L, Smith DW; The VATER association. Vertebral defects, anal atresia, T-E fistula with esophageal atresia, radial and renal dysplasia: a spectrum of associated defects. J Pediatr, 1973; 82(1): 104-107.