**Case Report**

**VACTERL association in the newborn**

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**ABSTRACT**

VACTERL association is a non-random association of birth (congenital) defects that affects multiple median and para-median structures. VACTERL association is a useful acronym to denote vertebral abnormalities (V), anal atresia (A), cardiac defects (C), tracheo-esophageal fistula (TE), renal or radial abnormalities (R), and limb abnormalities (L). At least 3 or more defects must be present to make a diagnosis of this condition. Most of these cases occur sporadically, although few cases with chromosomal abnormalities have been reported. Herein, we report a newborn who had most of the defects seen in VACTERL association.

**Introduction**

VACTERL association is a mnemonically useful acronym for a condition characterized by 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, tracheo-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 lobar defects, intestinal malrotation and genital anomalies [1]. Although diagnostic criteria vary, the incidence is estimated at approximately 1 in 10000 to 1 in 40000 live births [2]. In this report, we describe a newborn who was referred to us for the management of perinatal asphyxia and had most of the defects described in VACTERL association.

**Case Report**

A four hour old male baby, 2nd born to 3rd degree consanguinely married couple was referred to the outborn NICU of our institution with the complaints of the baby not having cried immediately after birth. The first child of the couple was 3 years old, alive and healthy. During this pregnancy the mother was registered and immunized. She underwent regular antenatal check-ups, however, she underwent only one obstetric scan in her last trimester of pregnancy which was told to be normal. Baby was delivered pre-term through vaginal route in a government hospital at Kollegal. There was no history of meconium stained amniotic fluid or prolonged labour. The baby did not cry immediately after birth, was resuscitated and referred to us for further management. On examination baby’s general condition was not satisfactory. He weighed 2kgs, had tachypnoea with increased work of breathing and signs of decompensated shock. spO₂ was maintained with 8-10lts of oxygen. He was euglycemic (GRBS – 90mg/dl), hypothermic and had poor cry and activity. There was microcephaly, hypertelorism, low set ears and high arched palate. There was persistent frothing from the mouth and we were unable to insert a nasogastric tube into the stomach. He had widely spaced nipples, symmetric hypoplasia of both radii with manuus valgus deformity and imperforate anus (Fig 1, 2 and 3). Systemic examination revealed a long systolic murmur at the lower left sternal margin. Moro’s and other newborn reflexes (sucking, rooting etc) were depressed. With the above findings an initial working diagnosis of syndromic baby with hypoxic ischemic encephalopathy was made and evaluated. His septic screen was negative, chest x-ray revealed coiling up of the nasogastric tube in the neck (suggestive of trachea-esophageal fistula) and hypoplastic thoracic vertebræ. Echocardiography
showed a perimembranous ventricular septal defect. USG abdomen, neurosonogram, ophthalmological examination and karyotyping were normal. Based on the phenotypic features a final diagnosis of VACTERL association with hypoxic ischemic encephalopathy was made. He was nursed in a thernomoteal environment and treated for hypoxic ischemic encephalopathy as per unit protocol. Parents were counselled regarding the congenital problems associated with the baby and the need for multiple and staged surgical procedures in an attempt to correct the anomalies. In view of the co-existent hypoxic ischemic encephalopathy and poor prognosis, parents decided to take the child home and he was discharged against medical advice.

Discussion

VACTERL association was earlier known as VATER association. The name was used for the first time in 1972 by American physicians David Weyhe Smith and Linda Quan [3]. VACTERL is a mnemonically useful acronym for a condition characterized by non-random association of specific birth (congenital) defects in structures derived from the embryonic mesoderm. Each letter in VACTERL represents the first letter of one of the more common findings (V-vertebral defects, A-anal atresia, C-cardiac defects, TE-tracheoesophageal fistula, R-renal anomalies, L-limb defects) seen in affected cases. Atleast 3 of the 7 criteria must be present before making a diagnosis of this condition. Only 1% of such cases present with the full range of anomalies [1].

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 1 in 10000 to 1 in 40000 live births [2]. The etiology is currently unknown, but is believed to be multifactorial [4]. The combination of VACTERL abnormalities can present with some known chromosomal abnormalities, including trisomy 13, 18, and 5p- syndrome. Intestinal deletion of long arm of chromosome 6 (6q13-15) and long arm of chromosome 13 have been reported in few cases [5]. 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 [6]. Children with VACTERL association are often born prematurely and with low birth weight. Affected children have multiple problems apparent at birth (congenital birth defects) and some of them could be observed on prenatal ultrasound. Additional characteristics of this association do not develop or are not apparent until later during life.

Vertebral anomalies are seen in 70% of these cases and consist of hypoplastic vertebral bodies, hemivertebrae, vertebral fusions and vertebral dysplasia. Most of these children have associated rib anomalies including supernumerary ribs, absent ribs, and fused ribs. In early life those anomalies rarely cause any symptoms, although the presence of these defects on a radiograph may alert the physician to other defects. Later in life, these vertebral anomalies may put the child at risk for developing scoliosis. While anal atresia or imperforate anus is seen in about 15%, cardiac defects are seen in about 25% of these cases. Among the cardiac defects reported, ventricular septal defects, atrial septal defects and tetrology of Fallot are most common. Oesophageal atresia with trachea-esophageal fistula is seen in about 70% of these cases. Renal anomalies are seen in 55% of these cases and include aplasia, hypoplasia, dysplasia, and agenesis of one or both kidneys. Absent or displaced thumbs, polydactyly, syndactyly, hypoplasia/aplasia of one or both radii are the most common limb defects seen in about 70% of these cases [7].

In addition to the above mentioned features, affected children may also exhibit less frequent abnormalities like failure to thrive, facial asymmetry (hemifacial microsomia), external ear malformations, lung lobation defects, intestinal malrotation and genital anomalies [8].

VACTERL association shows some phenotypic overlap with many other conditions including Feingold syndrome, CHARGE syndrome, 22q11 deletion syndrome, and Townes-Brock syndrome. 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].

Although children with VACTERL association have many problems, they can survive and become healthy. 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, otorhinolaryngologists and clinical geneticist in order to have a reasonable life expectancy [5].

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 [4].

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 [2,3].
Fig 1: Showing hypoplastic radii, manus valgus deformity and widely spaced nipples

Fig 2: Showing imperforate anus

Fig 3: X-ray of the baby showing hypoplastic radius
Conclusion

VACTERL association is a condition characterized by non-random association of specific birth defects involving multiple organ systems. Occurrence is usually sporadic. 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.

Conflict of interest statement

We declare that we have no conflict of interest.

References
