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Case Report

VATER Anomaly: A Rare Association

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ABSTRACT

VATER/VACTERL association is typically defined by the presence of at least three of the following congenital malformations: vertebral defect, anal atresia, cardiac defects, tracheoesophageal fistula, renal anomalies and limb anomalies. Most of these cases occur sporadically. Some cases with chromosomal abnormalities are reported. Here we report such rare newborn with VATER association.

Key words: Anal atresia, birth defects, sporadic, vertebral abnormalities, retrognathia.

INTRODUCTION

The VACTERL association (also VATER association) refers to the non-random co-occurrence of birth defects vertebral anomalies, anal atresia, Cardiac defects, tracheoesophageal fistula and/or esophageal atresia, Renal & Radial anomalies and Limb defects. Atleast 3 or more defects must be present to make a diagnosis of this condition. [1] In addition to these core components, patients may also have other congenital anomalies like hemifacialmicrosomia, external ear malformations, lung lobation defects, intestinal malrotation and genital anomalies. [2] The incidence is estimated at approximately 1 in 10000 to 1 in 40000 live births. [1] Although VACTERL or VATER are used as to represent same condition, VATER only has vertebral, anal, trachea-esophageal, and renal defects. Only 1% of such cases present with the full range of anomalies. [2]

CASE PRESENTATION

A 2 days female baby, product of a non-consanguineous marriage admitted in our SNCU with complaints of respiratory distress. The baby was born through normal vaginal delivery and not cried after birth. There was no h/o meconium stained liquor or prolonged labour. On 2nd day of life, baby developed respiratory distress and referred to our SNCU. On examination, baby was sick looking and sensorium was not good. Her weight was 1.8 kg. She was tachypneic with respiratory distress. SPO2 without O2 was 82%. Then maintained around 94% with O2. She was also having poor cry, reflexes and activity. On examination, the baby having microcephaly [figure 1], low set ear, retrognathia [figure 2, 3] and high arched palate [figure 5]. There was persistent frothing from mouth. We were unable to put NGT into stomach. On auscultation chest, there was b/l conducted sounds. There was a systolic murmur at lower left sternal border. Moros, rooting and suckling reflexes were poor. Based on these, our provisional diagnosis was syndromic baby with HIE. Chest X-ray revealed abnormal cervical vertebrae (unfused) with coiling up of NGT (suggestive of TEF) [figure 4]. Based on phenotypic features a

final diagnosis of VATER anomaly with HIE was made. She was managed as per HIE protocol. On 4th day of life, she became very sick and started gasping. Then baby became apneic and intubated and declared dead on 4th day of life.

Figure 1: showing microcephaly

Figure 2: showing low set ears, retrognatia

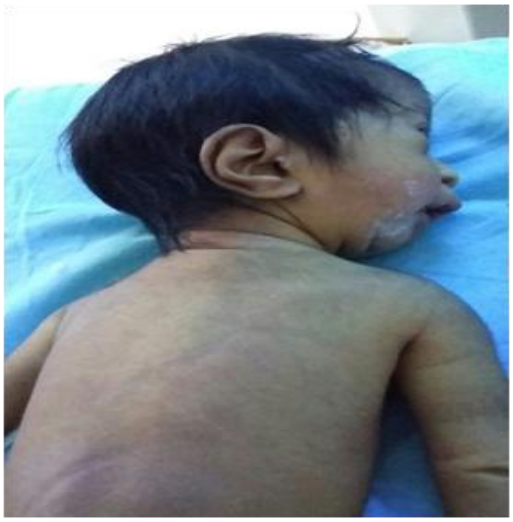

Figure 3: showing retrognathia, low set ears

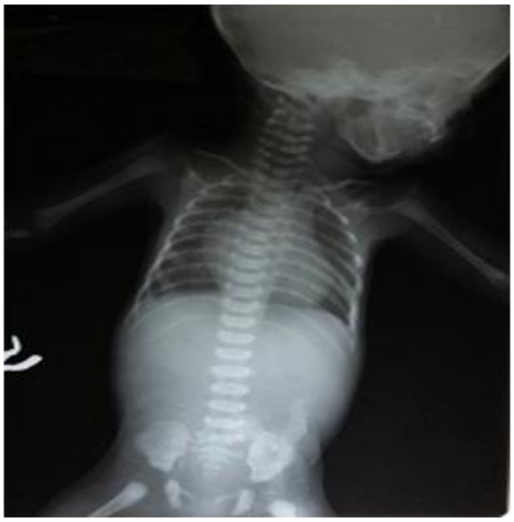

Figure 4: x ray showing unfused vertebrae, coiling of NG Tube suggestive of esophageal atresia

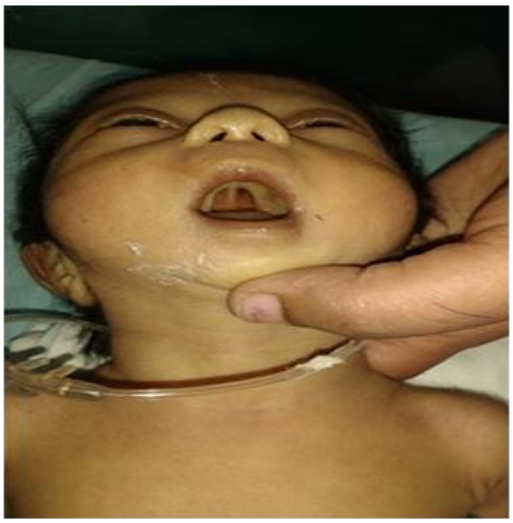

Figure 5: showing high arched palate

DISCUSSION

VATER is a non random association of specific birth (congenital) defect in structures derived from embryonic mesoderm. The name was used for the first time in 1972 by American physicians David Weyhe Smith and Linda Quan. [3] 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). Atleast 3 of the 7 criteria must be present before making a diagnosis of this condition. [1] Only 1% of such cases present with the full range of anomalies. [2] Although VACTERL or VATER are used as to represent same condition, VATER only has vertebral, anal, tracheoesophageal, and renal defects. The etiology is currently unknown, but is believed to be multifactorial. [7] The combination of VACTERL abnormalities can present with some chromosomal abnormalities. No specific genetic or chromosome problem has been identified with VACTERL association. VACTERL can be seen with some chromosomal defects such as Trisomy 18. Deletion of long arm of chromosome 6 (6q13-15) and long arm of chromosome 13 have been reported in few cases. [4] Baby with VATER anomaly is often born pre-maturely and with low birth weight.

Vertebral anomalies, or defects of the spinal column, usually consist of small (hypoplastic) vertebrae or hemivertebra where only one half of the bone is formed. About 80 percent of patients with VACTERL association will have vertebral anomalies. [5] In early life these 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 spinal column abnormalities may put the child at risk for developing scoliosis, or curvature of the spine. Anal atresia or imperforate anus is seen in about 15% cases. Cardiac defects are seen in about 25% of cases. The most common heart defects seen with VACTERL association are ventricular septal defect (VSD), atrial septal defects and tetralogy of Fallot. Less common defects are truncus arteriosus and transposition of the great arteries. [6] Esophageal atresia with tracheoesophageal fistula (TE fistula) is seen in about 70 percent of patients with VACTERL association, although it can frequently occur as an isolated defect. Renal (kidney) defects are seen in approximately 50 percent of patients with VACTERL association. In addition, up to 35 percent of patients with VACTERL association have a single umbilical artery. Limb defects occur in up to 70 percent of babies with VACTERL association and include a displaced or hypoplastic thumb, extra digits (polydactyly), fusion of digits (syndactyly) and forearm defects such as radial aplasia. [8] Features secondary to VACTERL components including single umbilical artery, ambiguous genitalia, abdominal wall defects, diaphragmatic hernia, and anomalies like intestinal and respiratory anomalies, and oligohydramnios sequence defects are frequent enough to be considered an extension of VACTERL. Many babies with VACTERL are born small and have difficulty with gaining weight. Babies with VACTERL association, however, do tend to have normal development and normal intelligence.

Diagnosis is mainly clinical and is based on the phenotypic features. Because the cause of VACTERL association is unknown, no laboratory test exists that can diagnose or rule out this condition. [8]

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. [4]

CONCLUSION

VATER anomaly is a rare association reported in very small no. of cases and involves multiple organ systems. Occurrence is usually sporadic. Some cases with chromosomal abnormalities are reported. Diagnosis is mainly clinical and is based on the phenotypic features. Multidisciplinary management is required with staged surgical approach being the mainstay of management.

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