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Case Report

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VACTERL ASSOCIATION IN A NEWBORN

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ABSTRACT

The acronym VACTERL describes the non-random co-occurrence of three of the following anomalies: Vertebral Anomalies (V), Anal (A), Cardiac (C), Tracheo-esophegeal fistula with or without Oesophageal atresia (TE), Renal (R) and lastly limb defects (L). Here, we report a newborn baby with VACTERL-type anomalies. The baby had dysmorphic features like short neck, low set ears and retrognathia. In addition to dysmorphic features the baby also had imperforate anus, congenital heart disease in the form of truncus arteriosus type 1 and left AV atresia with dominant right ventricle, Renal anomalies in the form of bilateral hydronephrosis and megaureter and lastly limb defects like polydactyly, valgus deformity of legs and rocker bottom feet.

KEY WORDS: VACTERL, Bilateral Hydronephrosis, Rocker Bottom Feet, Limb defects.

INTRODUCTION

The acronym VACTERL stands for Vertebral (V), Anal (A), Cardiac (C) anomalies, Tracheo-esophegeal fistula with or without Esophageal Atresia (TE), Renal (R) and lastly limb defects (L).[1] The constellation of anomalies seen in VACTERL association include vertebral (V) like hypoplastic or haemivertebrae , anal (A) like anal atresia, cardiac (C) like atrial septal defect, ventricular septal defect, tetralogy of fallot, truncus arteriosus and transposition of great arteries , trachea-esophageal fistula with or without oesophageal atresia (TE), renal (R) like posterior urethral valves, hydronephrosis and agenesis of kidneys and limb defects (L) which may include hypoplastic thumb, polydactyly, syndactyly and hypoplastic radius. The reported incidence of VACTERL association is 1: 10,000 to 1: 40000

depending upon the criteria used for establishing the diagnosis of VACTERL association.[2] The other anomalies which may be present in addition to these anomalies (non-vacterl) are single umbilical artery, genital defects and respiratory tract anomalies.[3] Though the exact etiology is not known, in some cases chromosomal anomalies like copy number variations.[4] and duplication in short arm of chromosome Y has been reported.[5]

CASE REPORT

A full term appropriate for gestational age male baby was delivered to a primigravida mother by normal delivery. In Antenatal anomaly scan the child was diagnosed to be having bilateral hydronephrosis with ventricular septal defect with single outflow tract. There was no family history of consanguinity or congenital birth defects. The baby cried immediately after birth with an Apgar score of 8/10 and 9/10 at 1 min and 5 min respectively. The weight of the baby was 2.3 kg (between 10th and 50th centiles); length (47 cm) and head circumference (32 cm) were both on 50th centiles. On general examination baby had short neck, low set ears, retrognathia, preaxial polydactyly, upper and lower limb anomalies (varus and valgus deformity with rocker bottom feet), umbilical hernia and imperforate anus.

Fig 1: Rocker bottom Foot seen in VACTERL association

Baby was admitted in NICU in view of multiple congenital anomalies. Baby was kept NBM and IV fluids and IV antibiotics were started. Child passed urine within 24 hours of birth. Baby was haemodynamically stable and was maintaining saturation. Routine investigations like CBC, Renal Function tests and thyroid profile was normal. Ophthalmological examination was done which revealed no abnormality. USG abdomen and 2 D ECHO was

also done. USG abdomen showed bilateral hydronephrosis and megaureter with urinary bladder detrusor hypertrophy with dilated rectum and normal looking anal canal which did not appear to be opening externally. 2 D ECHO was suggestive of complex cyanotic heart disease (truncus arteriosus type 1, left AV atresia with dominant right ventricle). Cranial Ultrasound was also normal. Taking into consideration the constellation of abnormalities a diagnosis of VACTERL association was made.

On the second day of life anoplasty was done. Baby tolerated the procedure well and postoperatively there were no complications. Baby was started on small nasogastric feeds after 24 hrs of anoplasty. Feeding was gradually increased and IV fluids were decreased accordingly. Baby tolerated feeds well and was on full feeds on D10. Baby was discharged on request of parents and was adviced to seek orthopaedics, plastic surgery and CVTS opinion for rocker bottom feet, syndactyly and feasibility of surgical correction of complex congenital heart disease.

DISCUSSION

VACTERL association as described above consist of Vertebral (V), Anal (A), Cardiac (C) anomalies, Tracheo-esophegeal fistula with or without Oesophegeal atresia(TE), Renal (R) and lastly limb defects (L). Because these defects occurred together more often than what one would expect by chance hence these anomalies were called association. There is no strict criteria that would constitute VACTERL association but majority of the scholars require that at least 3 defects should be present for labeling any patient to be having VACTERL association.[6] In addition to these abnormalities other defects may include hypothyroidism, cleft lip and cleft palate and some other abnormalities of respiratory and central nervous system. Though the definite etiology is not known many authors have reported chromosomal abnormalities in cases of VACTERL association. Faivre L et al concluded that "chromosomal breakage studies should be performed, not only in cases of VACTERL with hydrocephaly, but also in cases VACTERL with radial-ray anomalies and especially if the individual has additional FA associated manifestations such as skin pigmentation abnormalities, growth retardation, microcephaly, or microphthalmia".[7] It is more commonly seen in males. The preponderance in males may be explained by X-linked inheritance in some instances, sex-influenced expression, and mechanisms related to imprinting defects.[8] The Differential diagnosis of VACTERL association includes Baller-Gerold syndrome, CHARGE syndrome, Currarino syndrome, deletion 22q11.2 syndrome, Fanconi anemia,

Feingold syndrome, Fryns syndrome, MURCS association, oculo-auriculo-vertebral syndrome.[8]

Antenatal diagnosis can be difficult as many of the abnormalities involved are difficult to be picked up on antenatal scans. Some of the anomalies like renal agenesis or gross hydronephrosis can very well be picked up antenatally and may serve as a red flag sign for the presence of other anomalies.[9] The management of patients with VACTERL association requires immediate intervention in cases of anal atresia, trachea-esophegeal fistula and some cardiac defects. This immediate surgical or medical intervention must be followed by long term medical management for various associated anomalies. The prognosis is relatively better once the surgical correction is achieved. Absence of neurocognitive defects and a normal IQ means with proper management of immediate surgical emergencies and prolonged medical care the patients with VACTERL association can be expected to have a satisfactory outcome.[10]

CONCLUSION

Though VACTERL association is rare, detection of any congenital anomaly, on antenatal scan or postnatally, which is a part of VACTERL association must alert a pediatrician to look for additional anomalies so that timely intervention can be done and a better outcome is expected.

Conflict Of Interest: None

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