Full Spectrum of VACTERL in New Born

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MJAFI 2008; 64 : 84-85

Key Words : Neonate; Congenital anomaly

Introduction

The VACTERL (vertebral anomalies, anal atresia, cardiovascular malformations, tracheo-oesophageal fistula, renal and limb anomalies) association is a non random pattern of defects occurring together and includes at least three of the above cardinal features. The finding of the full VACTERL spectrum has however, not been reported. We report the case of a newborn who manifested the complete VACTERL association along with other dysmorphic features.

Case Report

A preterm baby weighing 1250 grams was born at 35 week gestation to a 25 year old second gravida mother with an uncomplicated antenatal period. There was no history of exposure to any known teratogen. The antenatal ultrasound examinations (done at 30 and 34 weeks) showed double bubble appearance in abdomen. The baby was severely asphyxiated at birth. Endotracheal intubation and resuscitation was carried out. The anomalies were low set ears, epicanthal folds, high arched palate, oesophageal atresia (inability to pass red rubber catheter in oesophagus), absent right radius bone (radial dysplasia), single bone in the left lower limb, left foot not formed, absent anal opening, thoracic hemivertebra, lower limbs fused at thighs and genitalia not well formed (ambiguous). A single umbilical artery was noted at the time of cord ligation (Fig. 1). The head circumference was 29 cm and length was 39 cm. Baby died after 17 hours of birth. The autopsy findings were small dysplastic kidneys, multiple intestinal atresias and transposition of great vessels.

Discussion

The acronym, VATER was adopted in 1972 to describe a cluster of vertebral, anal, tracheo-oesophageal and radial malformations. This non random association has a birth prevalence varying from 1:3,500 to 1.6:10,000. Heart, renal and other limb malformations have also been reported adding the letters C, R and L to the acronym [1]. Babies who have been diagnosed as having VACTERL association usually have at least three or

more of these anomalies but the entire spectrum has not been reported earlier.

Majority of these patients have congenital heart disease, with ventricular septal defect (VSD), atrial septal defects and tetralogy of Fallot being the commonest. Vertebral anomalies usually consisting of hypoplastic vertebrae or hemivertebra are seen in about 70 percent of patients. Anal atresia or imperforate anus is seen in about 55 percent and oesophageal atresia with tracheo-oesophageal fistula in 70 percent of patients. Fifteen to 33 percent of patients with tracheo-oesophageal fistulas will also have congenital heart disease.

Renal defects are seen in half the patients with malformation of one or both kidneys or obstructive uropathy. In addition, up to 35 percent of patients with VACTERL

Fig. 1 : Full spectrum of VACTERL in a single newborn : low set of ears, anorectal malformation, radial dysplasia, single umbilical artery, oesophageal atresia with tracheo-oesophageal fistula, lumbar hemivertebra and webbed thighs

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association have a single umbilical artery, which can often be associated with kidney or urologic problems. Limb defects seen in up to 70 percent of babies include absent or displaced thumbs, polydactyly, syndactyly and forearm defects. Babies with limb defects on both sides tend to have kidney or urologic defects on both sides, while babies with limb defects on only one side of the body tend to have kidney problems on the same side. VACTERL association carries a significant risk of mortality.

A defect in blastogenesis has been suggested as a possible aetiology of this malformation. Martinez-Frias et al proposed that combinations of anomalies of blastogenetic origin, such as VATER/VACTERL should be considered and called “polytopic field defects” instead of the generic term “association” [2,3].

Defects of practically every organ system have been reported in association with VACTERL [4]. Feingold syndrome, CHARGE syndrome, 22q11 deletion syndrome, Townes-Brocks syndrome, and Pallister-Hall syndrome show some phenotypic overlap with the VACTERL association, and most series of patients ascertained by epidemiological means are not divided into subgroups according to confirmed or possible syndromic diagnoses [5].

Individuals with the VACTERL association do not typically have facial dysmorphic features, learning disability or abnormalities of growth, including head circumference. Many babies with VACTERL are born small and have difficulty with gaining weight although development and intelligence in survivors is usually normal.

Conflicts of Interest

None identified

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