

Eyelid Coloboma in a Newborn with VACTERL Association

Timilsina M¹, Gauchan E², Koirala DP³

Abstract

VACTERL is used to denote vertebral abnormalities (V), anal atresia (A), cardiac defects (C), tracheoesophageal fistula (TE), renal or radial abnormalities (R), and limb abnormalities (L). This is a case report of a neonate delivered at Manipal Teaching Hospital, Pokhara with features suggestive of VACTERL association.

Key words: Eyelid Coloboma, Newborn, VACTERL association

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Introduction

Described for the first time in 1972 by David Weyhe Smith and Linda Quan¹, the acronym VATER describes the components: Vertebral defects, Anal atresia, Tracheoesophageal fistula with oesophageal atresia, and Radial and Renal dysplasia. Kaufman (1973) and Nora and Nora (1975) subsequently added "C" for cardiac defects and "L" for limb defects². Patients may also have hemi facial microsomia, external ear malformations, lung lobation defects, intestinal malrotation and genital anomalies³.

The Case

A new-born delivered via Caesarean Section for pregnancy-induced hypertension was shifted to NICU immediately after birth for delayed cry and multiple congenital anomalies. The baby was born to a 29-year mother via non-consanguineous marriage at 40 weeks of gestation. The previous child is a normal three year old male. The mother did not receive any antenatal check-ups. There was no history of infections, drug intake, radiation exposure or any family history of congenital anomalies. The baby had APGAR score of 3 at 1 minute and required immediate intubation and cardiopulmonary resuscitation following which the baby's condition improved, APGAR score was 8 at 5 minute and saturation was maintained without oxygen. He weighed 2.4 kg (<10 percentile), 49 cm in length (50th percentile) and had occipito-frontal circumference of 30 cm (<10 percentile) and was euglycaemic (GRBS-50mg/dl). There was persistent frothing from the mouth including occasional bilious vomiting, with inability to insert nasogastric tube into the stomach. He had coloboma of the right upper eyelid (Figure 1), skin tag over the medial canthus of the right eye, preaxial polydactyly of the right upper limb (Figure 2), undescended testis on the left side and imperforate anus (Figure 3). Neonatal reflexes including Moro's reflex were depressed. Systemic examination revealed a pansystolic murmur of grade 3/6 at the lower

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left sternal border. His initial septic screen was negative, Chest X-ray revealed coiling of the nasogastric tube at the level of fifth thoracic vertebrae (Figure 4), and hemivertebrae of the tenth thoracic vertebrae (Figure 5). The right kidney could not be visualized by ultrasound scan and there was mild left hydro-uretero-nephrosis. Ophthalmic examination did not reveal any additional anomalies. The patient was nursed in a thermo-neutral environment with maintenance IV fluids for three days. The patient passed urine stained with meconium suggesting recto-urethral/recto-vesical fistula. The parents were advised for further investigation including echocardiogram which they refused. They were properly counselled regarding congenital anomalies and need for multiple surgeries and intensive therapy for the management of the illness. Because of multiple anomalies and poor outcome of the illness they decided to take the child home against medical advice.



Fig 1: Eyelid Coloboma of right with small skin tag



Fig 2: Polydactyly of right hand



Fig 3: Imperforate Anus

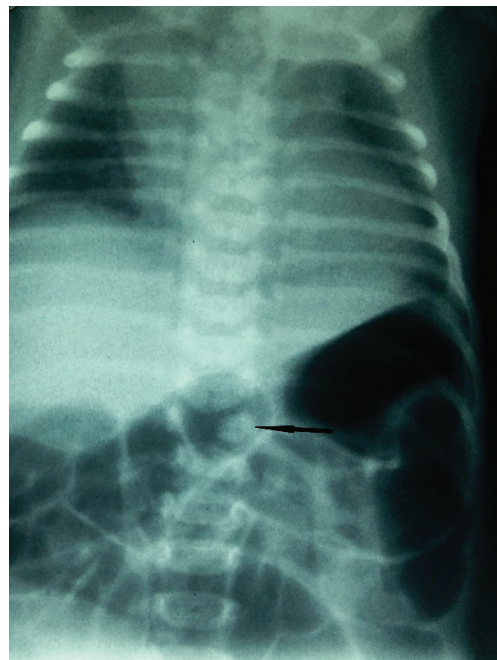


Fig 4: Hemivertebrae of T10

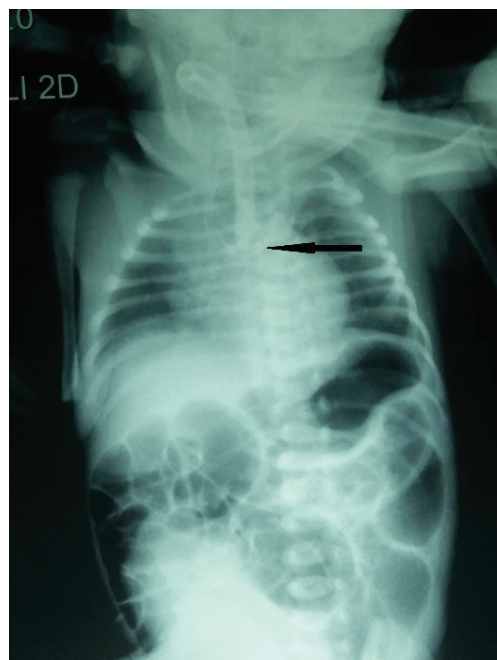


Fig 5: Coiling of the NG tube at level of T5

Discussion

The incidence is estimated to be approximately 1 in 10000 to 1 in 40000 live births⁴. The aetiology is unknown and is found to occur sporadically in an otherwise normal family. It is sometimes associated with Infants of Diabetic Mother and Fanconi's Anemia⁵.

In this primary polytopic developmental field defect^{5,6}, there is disruption in differentiating mesoderm in first 4-5 weeks causing defect in the blastogenesis⁸. Defective Sonic hedgehog pathway signalling during human embryogenesis is also implicated⁹.

Individuals with VACTERL association have the following anomalies with varying incidence⁴:

1. Vertebral anomalies usually hypoplastic vertebrae or hemi-vertebra are seen in 70% of the cases.
2. Anal atresia with or without fistula and imperforate anus are seen in 80% of patients.
3. Cardiac defects comprising of ventricular septal defects, atrial septal defects, Tetralogy of Fallot, truncus arteriosus and transposition of the great arteries are seen in 53% of cases.
4. Tracheoesophageal fistula with oesophageal atresia are seen in 70% patients.
5. Renal anomaly malformation of one or both kidneys or obstructive uropathy are reported in about 53% of patients.
6. Limb anomalies including thumb or radial hypoplasia, pre-axial polydactyly, syndactyly, absent or displaced thumbs, and leg defects are seen in 63% of patients.
7. Single umbilical artery is found in 35% of patients.

Other defects associated with VACTERL are facial asymmetry (hemi-facial microsomia), external ear malformations, lung lobation defects, intestinal malrotation and genital anomalies, abdominal wall defects, diaphragmatic hernia, prenatal/postnatal growth deficiency, laryngeal stenosis, bronchial anomalies, large fontanels, defect of the lower limb, rib anomaly, occult spinal dysraphia with tethered cord, respiratory

anomalies and oligohydramnios sequence defects^{5,10}.

The differential diagnosis includes Baller Gerold syndrome, CHARGE syndrome, Currarino syndrome, Deletion 22q11.2 syndrome, Fanconi anemia, Feingold syndrome, Fryns syndrome, MURCS association, Oculo-auriculo-vertebral syndrome, Opitz G/BBB syndrome, Pallister–Hall syndrome, Townes–Brocks syndrome and VACTERL with hydrocephalus⁴.

There should be at least three out of the seven core features, to diagnose a case as VACTERL association¹¹. But only 1.0% of such cases present the full range of anomalies². Ultrasonography and MRI can visualize some of the characteristic findings of this condition prenatally¹².

Management requires a multidisciplinary approach including paediatrician, cardiologist, urologist, orthopaedic surgeons, oto-rhino-laryngologists and clinical geneticist in order to have a reasonable life expectancy¹³.

According to a 10 year follow-up of infants with VACTERL, the estimated mortality rate was 24%, mainly due to cardiovascular abnormalities¹⁴. Morbidity is significant following repair of trachea-oesophageal abnormalities. Anorectal and vertebral anomalies are associated with variable but poor functional outcomes in later life¹⁵.

Prognosis depends on the severity of anomalies. Prognosis in resource restricted nations like ours is very poor.

Conclusion

VACTERL is a non-random sporadic association causing multiple congenital defects of varying severity and presentation. Staged surgical management of various defects is the primary modality of treatment. There is no definite tool for prenatal diagnosis or confirmation of the association. Because of lack of specific management prognosis is poor in the affected infant. However recurrence is rare in subsequent pregnancies.

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