Case Report

# A RARE CASE OF VACTERL ASSOCIATION AT THE FEDERAL MEDICAL CENTRE, YENAGOA, BAYELSA STATE, NIGERIA.

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## Abstract

**Background:** VACTERL association is a condition comprising multisystemic congenital malformations, causing severe physical disability in affected individuals and is usually a great burden to the family and challenging for the Paediatrician. VACTERL/VACTER is characterized by presence of vertebral defects, anal atresia, cardiac defects, trachea-oesophageal fistula, renal anomalies and limb abnormalities.

**Case presentation:** A live male baby was delivered at the labour ward of the Federal Medical Centre, Yenagoa, following preterm pre-labour rupture of foetal membranes at 30 weeks' gestational age with multiple congenital malformations. The neonate was immediately transferred to the special care baby unit (SCBU) of the facility for expert care. Baby died within four hours of life.

**Conclusion:** VACTERL association is a very rare congenital anomaly. Aetiology is unknown and diagnosis is made on clinical picture. Management is quite challenging and multidisciplinary approach is needed to optimize neonatal survival.

**Keywords:** VACTERL association, Congenital malformations, Multidisciplinary.

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## INTRODUCTION

Congenital anomalies are important causes of infant and childhood deaths, chronic illnesses and disability. Congenital anomalies or birth defects are structural, behavioural, functional and metabolic disorders that occur during intrauterine life and can be identified during the antenatal period, at birth or later in infancy.[1,2]

Prematurity, birth asphyxia and infections are the major causes of adverse neonatal outcomes. Congenital anomalies also contribute significantly to neonatal morbidity and mortality.[1,2]

Major Congenital abnormalities occur in 2-3% of live births and 20-30% of still births.[1,3] One of such major congenital anomalies involving multiple organs/system is VACTERL/VACTER association. VACTERL association is a condition comprising multisystemic congenital malformations, causing severe physical disability in affected individuals, and is usually a great burden to the family and challenging for the Paediatrician. The components of VACTERL or VACTER association are vertebral anomalies, anorectal malformations, cardiovascular anomalies, tracheo-oesophageal fistula, renal anomalies and limb defects.[3]

## CASE PRESENTATION

A live male baby was delivered vaginally at the labour ward of the Federal Medical Centre, Yenagoa, following preterm prelabour rupture of foetal membranes at 30 weeks' gestational age with multiple congenital malformations. APGAR scores were $4^1$, $6^5$ and birth weight 2.2kg. The neonate was immediately transferred to the special care baby unit (SCBU) of the facility for expert care.

The mother was a 35-yearr-old trader with primary level of education who resided in Akaba town, in Bayelsa State, Nigeria. She was $P1^{+1}$ (not alive). Pregnancy was booked for antenatal care at 13 weeks' gestational age, and was uneventful until delivery. She could not remember her booking parameters and results of her booking investigations. She did not do any ultrasound scan in this pregnancy. She received 2 doses of tetanus toxoid and 2 doses of sulphadoxine/pyrimethamine for intermittent preventive treatment for malaria. She was regular on her visits, and did not take any other medications apart from her routine haematinics. There was no family member that had similar features or had a child with similar features. She had one premarital termination of pregnancy at a private clinic through manual vacuum aspiration at 8 weeks' gestation. She did not have any known chronic medical condition.

Musculoskeletal examination of the baby revealed vertebral defects, limb defects and imperforate anus **[Figure 1].** Baby however, died within four hours of life. The parents were counselled for autopsy but declined.

**Figure 1: Neonate with VACTERL association.**

## DISCUSSION

VACTERL is a mnemonically useful acronym for the non-random association of vertebral defects (V), Anal atresia (A), cardiac malformations (C), tracheo-oesophageal fistula (TE), renal anomalies (R), limb defects (L). The aetiology of VACTERL association is not known, and its occurrence is mainly sporadic. Inherited factors and causal heterogenicity have been implicated in the aetiology of VACTERL association.[4,5,6]

It has been shown to be commoner in middle- and low-income earners as in the case presented.[7] She did not have any family history of such anomalies. Diagnosis of VACTERL association is based on the clinical picture at birth and presently there is no general consensus on diagnostic criteria but most clinicians and researchers require the presence of at least 3 component features for diagnosis.[7]

Clinical- or laboratory-based evidence are important in making diagnosis of this condition.[7] The neonate had visible vertebral defects (as evidenced by curvature of the spine) imperforate anus (which is an anorectal defect) and visible limb abnormalities. These were enough to make the diagnosis clinically, though laboratory and radiological investigation were not done prior to the death of the neonate.

Diagnostic criteria for VACTERL association vary. The incidence is about 1/10,000 – 1/40,000 live births.[7,8,9] VACTERL association does not have any strong evidence for higher incidence in particular regions of the world or in specific ethnic groups.[7,8,9] Some but not all studies have suggested that the condition is more common in males as in the case presented.[7,8,9]

The differential diagnosis of VACTERL is broad and includes a number of conditions for which genetic testing is available.[7,8,9] These conditions include Baller-Gerold syndrome, CHARGE syndrome, Currarino syndrome, DiGeorge syndrome, Fanconi Anaemia, Feingold syndrome.[7,8,9]

The requested investigations were yet to be done before the death of the neonate. Antenatal diagnosis is quite challenging as certain components can be difficult to ascertain prior to birth. However, certain clues can suggest VACTERL type anomalies such as

polyhydramnios, lack of gastric bubble due to tracheo-oesophageal fistula and a dilated colon due to imperforate anus.[8] It is important to emphasize that the discovery of a single umbilical artery may be the first clue to diagnosis[8]. All these findings can be detected by high resolution ultrasound scan. The mother of the neonate did not do any ultrasound during pregnancy.

The main treatment modality for patients with VACTERL association is surgery, which involves surgical correction of certain congenital anomalies like anal atresia, cardiac malformations, and tracheo-oesophageal fistula during the post-partum period. This is accompanied by management of the long-term medical co-morbidities of the congenital malformations.[8,9] The neonate died within four hours of life and the parents refused autopsy which may have given further support to the clinical diagnosis. Many people in this part of the world have aversion to autopsy for various reasons. An example is a case report in this centre, where a neonate with congenital malformations died within hours of delivery; and the parents refused autopsy despite profuse and adequate counselling.[10]

The prognosis of VACTERL association is generally good if optimal correction of anomalies is achievable, although some patients will continue to be affected by their congenital malformations throughout life.[11,12]

## CONCLUSION

VACTERL association is a very rare congenital anomaly. Aetiology is unknown and diagnosis is made on clinical picture. Management is quite challenging and multidisciplinary approach is needed to optimize neonatal survival.

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