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Cardiac, Renal and Gastrointestinal Anomalies in a Neonate: Could this be VACTERL Association? A Case Report

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Authors' contributions

This work was carried out in collaboration among all authors. Authors UI and COD designed the study and wrote the first draft of the manuscript. Authors OO, IITO and SO managed the literature searches. All authors read and approved the final manuscript.

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Case Study

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ABSTRACT

Introduction: VACTERL association is a rare congenital defect. It is a constellation of the involvement of six systems which comprise of vertebral defects, anal atresia, cardiac defects, trachea-oesophageal fistula, renal anomalies and limb abnormalities. At least three systems should be involved before VACTERL is suspected and in many cases, it is difficult to differentiate it from other multiple congenital anomalies.
Case Report: We present a day old preterm neonate who presented with respiratory distress and abdominal distension at birth. Late pregnancy ultrasound done at 34 weeks gestation showed a singleton fetus with gross ascites, dilated urinary bladder and bilateral calyceal dilatation. On examination, he had hyper-plantar flexion of the left ankle joint, ascites and a loud systolic murmur.

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Abdominal scan showed bilateral renal stones with medullary sponge kidneys, gaseous distension of the bowels and massive ascites. Transthoracic echocardiography showed a 10 mm ostium secundum atrial septal defect, 6 mm perimembranous ventricular septal defect and a 3 mm patent ductus arteriosus. Micturating cystourography showed a dilated posterior urethra with an appearance of a ring lucent filling defect at the membranous urethra and an irregular and beading distal urethra which was suggestive of posterior urethral valves (diaphragmatic type).
Conclusion: VACTERL association occurs sporadically in most cases and presentation is varied depending on the degree of systemic affectation. Our patient presented with a constellation of congenital defects which could all fit into the criteria for VACTERL association, however because of the lack of genetic testing, it is difficult to determine if this is just a chance occurrence of multiple congenital anomalies.

Keywords: VACTERL association; renal anomalies; cardiac anomalies; gastrointestinal anomalies.

1. INTRODUCTION

VACTERL association is a rare congenital defect. It is a cluster of congenital malformations with multi-systemic involvement and comprises of at least three of the following: vertebral defects, anal atresia, cardiac defects, tracheo-esophageal fistula, renal anomalies, and limb abnormalities [1]. Less than 1% of cases feature all six malformations [2,3]. It has an incidence of 1 in 10,000 to 1 in 40,000 live-birth infants and most cases occur sporadically though familial occurrences have been described [4,5]. The involvement of multiple systems has necessitated the acronym- VACTERL for appropriate identification of the defect, however other congenital defects could bear similarities to it. We report the case of a new born with a constellation of cardiac, gastrointestinal, musculoskeletal and renal abnormalities suggestive of VACTERL association.

2. PRESENTATION OF THE CASE

Baby C presented at the Special Care Baby Unit of the Federal Medical Centre Yenagoa at 30 minutes of life in severe respiratory distress. He had been delivered via Emergency caesarean section at a gestational age of 34 weeks +5 days to a 27 year old unbooked primigravida on account of prolonged preterm rupture of membranes. Apgar scores were 6 at 1 minute and 7 at 5 minutes respectively and he had been resuscitated by airway suctioning, minimal stimulation and intranasal oxygen via face mask. He weighed 3 kg at birth.

The child was the product of a non-consanguineous union. The pregnancy had been supervised at a government hospital from 16 weeks of gestation. The mothers' HIV I&II serology, HBsAg and HVC were negative and VDRL was non-reactive. She had been on routine haematinics and cod liver oil from the first month of pregnancy and there was no history of use of local concoctions or alcohol or exposure to radiation. Late ultrasound done at 34 weeks gestation showed a singleton fetus with gross ascites, dilated urinary bladder and bilateral calyceal dilatation.

On examination at delivery, he was acutely ill and in severe respiratory distress with no obvious dysmorphic features apart from hyper-plantar flexion of the left ankle joint. He was dyspneic with a respiratory rate of 86 cycles per minute and had transmitted sounds bilaterally. His oxygen saturation ranged from 88% in room air to 92% on 100% oxygen. His heart rate was 156 beats per minute and regular, with the apex beat located on the 4th left intercostal space and a grade 4/6 systolic murmur located at the left sternal border. His abdomen was massively distended with visible abdominal veins, and abdominal girth measured 6cm from the xiphisternum was 39.2 cm. His abdominal organs were difficult to palpate and the bowel sounds were hyperactive. He had normal external male genitalia and his anus was patent. He was conscious and had a good cry, patent fontanel and an occipitofrontal circumference of 32 cm with normal tone and reflexes.

Chest X-ray done showed a fairly homogenous opacity in the left lung field, obscuring the left cardiac border and a mid-clavicular fracture on the left. On echocardiography, there was a large unrestrictive 10mm atrial septal defect 6mm perimembranous ventricular septal defect and 4mm patent ductus arterisous. Abdominopelvic scan showed that the kidneys were normal in size, cortical echogenicity and corticomedullary differentiation but had multiple foci of granular hyper-echogenicity in the calyces with mild hydronephrosis and massive ascites suggestive of bilateral renal stones with medullary sponge

kidneys. The liver and spleen appeared normal but the pancreas was not visualized. Abdominal X-ray revealed gas filled bowel loops with no air-fluid levels suggestive of Meteorism. Micturating cystourography showed a dilated posterior urethra with an appearance of a ring lucent filling defect at the membranous urethra and an irregular and beading distal urethra. An impression of Posterior urethral valves - Diaphragmatic type was made.

A diagnosis of a newborn with multiple congenital anomalies in heart failure with neonatal septicaemia and left clavicular fracture from birth trauma was made and the baby was placed on intravenous antibiotics, intravenous fluids, oral frusemide and oral spironolactone. He was co-managed with the paediatric surgical and orthopaedic team and was scheduled for surgery when his condition became stable. Electrolyte derangements were corrected and he was transfused with blood on account of anaemia with haemoglobin level ranging from 10.5 g/dl to 12.3g/dl. His condition however remained poor throughout admission despite intervention and he developed apnea on the 6th day of life and subsequently died.

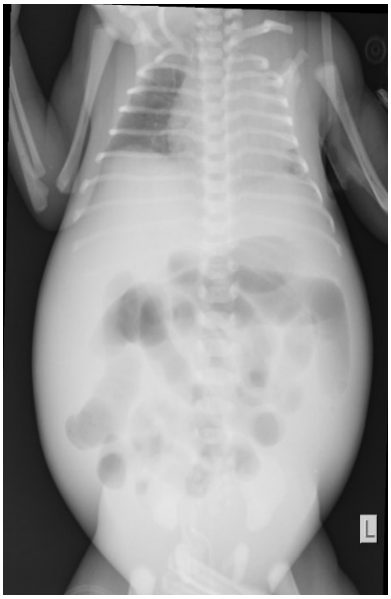

Fig. 1. Chest and abdominal radiograph showing cardiomegaly, reduction of the left lung field with ipsilateral shift of the mediastinum and some rib crowding suggesting some collapse and fracture of the mid-portion of the left clavicle. There is ascites, enlarged liver and gaseous distension of the small intestines

3. DISCUSSION

VATER association was first described in 1972 by Quan and Smith and was regarded as a non –random co-occurrence of the following component features of vertebral anomalies (V), anal atresia (A), tracheo-esophageal fistula and /or esophageal atresia (TE), radial dysplasia (R) [6]. A year later, same authors linked the letter "R" not only to radial dysplasia but also to renal anomalies [6]. In 1974, Tentamy and Miller added cardiac (C) and limb defects (L), and the acronym was revised to VACTERL association [7]. Several extension of the clinical definition were proposed in the following years to include vascular anomalies (V), auricular anomalies (A) and rib anomalies (R) [8]. The VACTERL association occurs sporadically in most cases though the involvement of genetic factors is suggested by reports of familial occurrence of some of the characteristic features among first-degree relatives of affected individuals, high concordance rates among monozygotic twins, chromosomal aberrations or single gene mutations in individuals with the VACTERL phenotype [6].

The presence of clinical features as was found in our patient which involved multiple systems qualifies for a clinical diagnosis of VACTERL involving the cardiac, digestive, renal and musculoskeletal system. There are varied presentation of the systemic affectation. Despite the fact that vertebral anomalies and trachea-esophageal fistula/esophageal anomalies have been shown to occur in 60-70% of affected patients [9], our patient had neither. Anal atresia which is also described as a common component of the association was not reported in our patient though he did have atretic distal bowel loops with proximal bowel gaseous distension. Similar features have been documented in literature where duodenal atresia rather than trachea-esophageal atresia and anorectal malformation was reported in patients with the VACTERL association [10,11].

Our patient also presented with involvement of the renal system- bilateral kidney stones, hydronephrosis, medullary sponge kidneys and posterior urethral valves. This is in keeping with the findings by Cunningham et al. [12] who reported that the most frequent renal manifestation is his VACTERL cohort was vesicoureteric reflux in conjunction with a structural anomaly. Isolated vesicoureteric and hydronephrosis are also common in patients with

VACTERL [12], which were similarly found in our patient. The various types of renal anomalies seen has been attributed to be dependent on the level and nature of the underlying genetic lesion [12]. Cunningham et al. [13] in their study found out that 67% of the children that met the criteria for VACTERL association had congenital heart disease (CHD) of which ventricular septal defects (VSD), atrial septal defects (ASD) and Tetralogy of Fallot (TOF) were the commonest seen, This is consistent with our findings of VSD and ASD though PDA has not been commonly reported in literature. The authors also noted that the presence of CHD did not correlate with the presence of trachea-esophageal fistula or anorectal malformation [13], a finding similarly observed in our patient.

Reports have shown that newborns with VACTERL association and ipsilateral renal disorders have the same side limb defects [14]. This contracts with our case who had bilateral affectation of the kidneys and unilateral affectation of the left lower limb. In addition to the lower limb defects, our patient was also noted to have a left clavicular fracture but this was however thought to be due to birth trauma rather than a congenital anomaly. Upper limb anomalies such as thumb aplasia/hypoplasia have been described as the more common limb deformities observed in the VACTERL association [1]. However, there have been reported cases of VACTERL with lower limb affectation such as club foot and calcaeneus valgus [15] which fits into the presentation of the unilateral left ankle affectation that was seen in our patient.

4. CONCLUSION

VACTERL association is a rare congenital defect with variable presentation depending on the degree of systemic involvement. Our patient presented with a constellation of congenital defects which could all fit into the criteria for VACTERL association, however with the lack of genetic testing it is difficult to differentiate this from a coincidental occurrence of a cluster of multiple congenital anomalies.Though the diagnosis cannot positively be confirmed as being VACTERL, it remains the strongest possibility.

CONSENT

As per international standard or university standard, patient's consent has been collected and preserved by the authors.

ETHICAL APPROVAL

It is not applicable.

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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