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Case Report

Association of VACTERL with truncus arteriosus, left-sided aortic arch, hypoplastic pulmonary arteries, and severe radial axis defect

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ABSTRACT

Background: VACTERL association is usually a sporadic disorder, the possible etiologies of which have been proposed as familial as well as multiple genetic and environmental factors. VACTERL association usually consists of at least three of the core features of vertebral defects, anal atresia, cardiac defects, tracheoesophageal fistula, renal anomalies, and limb abnormalities. Vertebral anomalies, cardiac malformations, renal anomalies, and limb malformations have been reported to occur in 60-80%, 40-80%, 50-80%, and 40-50% of the patients, respectively. Among the cardiac anomalies, ventricular septal defect is the most prevalent one. Truncus arteriosus is a rarely associated defect. The radial anomalies are the most classic limb defects with different severity levels (types I-IV). Although radial axis defect usually accompanies this association, complete absence of radius is reported only in one third of the cases.

Case report: In this case study, we reported a neonate having this association consisting of severe cyanotic congenital heart defect (truncus arteriosus) and unilateral type IV radial aplasia.

Conclusion: VACTERL association components can have various new findings.

Keywords: Neonate, Truncus arteriosus, VACTERL

Introduction

VACTERL association is a rare disorder with the incidence rate of 1 newborn per 10,000 to 40,000 cases that affects many body systems. This disorder consists of vertebral defects, anal atresia, cardiac defects, tracheoesophageal fistula, renal anomalies, and limb abnormalities (1-4). The patient having at least three of these characteristic features are diagnosed with VACTERL association. Ventricular septal defect is the most prevalent cardiac disorder. The complete absent radius is reported in one third of the cases. Herein, we presented a neonate suffering from VACTERL association consisting of severe cyanotic congenital heart defect and unilateral radial aplasia.

Case report

Our case was a male neonate born to a 22-year-old primigravida by vaginal delivery, who was referred at four hours of life due to respiratory distress and cyanosis. His birth weight was 2,900 g. Upon admission, the peripheral capillary oxygen saturation was 66% in ambient air. Arterial blood gas analysis revealed severe hypoxemia (PaO_2: 38mmHg) and compensated metabolic acidosis. He was intubated and put on SIMV mode of ventilation.

Further examination revealed narrow palpebral

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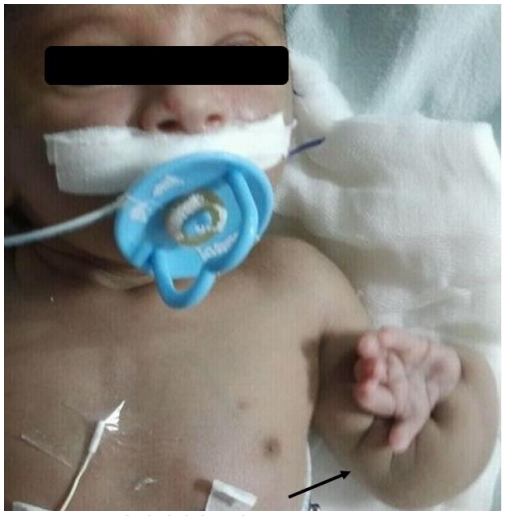

Figure 1. Radial club hand

fissure, broad nasal root, low-set small ears, radial club hand, small thumb, and simian crease on the left side (Figure 1), sandal gap, imperforate anus, cardiac apex in the left 4th intercostal space, and grade 3/6 short systolic murmur at the left lower sternal border. The results of the other systemic examinations were within the normal limits.

The chest X-ray revealed cardiomegaly, oligemic lung fields, and a defect in the sixth thoracic vertebra (hemivertebra). Furthermore, the X-ray of the left arm showed radial aplasia (type IV). Echocardiography demonstrated single great artery (type 2), large ventricular septal defect (VSD), secundum atrial septal defect (ASD, 3mm), left-sided aortic arch, and hypoplastic pulmonary arteries bilaterally. In addition, the abdominal ultrasound revealed dilated rectum with air-fluid levels till sacrococcygeal joint and normal kidneys.

Neurosonogram was normal. The neonate had episodes of hypoglycemia, which required high glucose infusion rates (up to 12mg/kg/min) to achieve normoglycemia, hypocalcemia requiring calcium supplementation, and hyponatremia within the initial five days. There was a transient renal failure, which was spontaneously resolved by 10 days. Karyotyping did not show chromosome 22q deletion.

Further tests, such as fluorescent in situ hybridization or array comparative genomic hybridization, were not performed in this case due to financial constraints. Diuretic (frusemide, 1mg/kg body weight, once daily) was administered to control congestive heart failure. Anoplasty was performed under anesthesia. Tube feeds were started on day four of life and graded up. Gradually, direct breastfeeding was established, and the neonate was discharged. During the one month follow-up, the newborn was found to have cyanosis and inadequate weight gain. The cardiac surgery was planned at later date since the surgery was carried out only at the selected centers of the region, and the parents wanted time to arrange finance.

Discussion

VACTERL association is usually sporadic in occurrence; however, it is caused by familial as well as multiple genetic and environmental factors. The association is characterized by the presence of at least three of the six core congenital malformations, including vertebral defects, anal atresia, cardiac defects, tracheoesophageal fistula, renal anomalies, and limb abnormalities (1).

Vertebral anomalies have been reported in 60-80% of the patients. They include segmentation defects, such as hemivertebrae, butterfly vertebrae (wedge fracture), vertebral fusions, supernumerary or absent vertebrae, and other forms of vertebral dysplasia. Vertebral defects are often accompanied by rib anomalies (1-4). The present case had hemivertebra of thoracic segment six. Imperforate anus occurs as a part of anorectal malformation in 55-90% of the cases. Anal stenosis may also be considered as a component of the association. In addition, genitourinary anomalies may be observed in 25% of the patients. The present case had anal atresia requiring anoplasty in the immediate neonatal period.

Cardiac malformations have been described in highly variable percentages. About 40-80% of the patients with VACTERL association have congenital heart defects. The most common heart defects include ventricular septal defect, atrial septal defect, and tetralogy of Fallot (2). The present case had a rare cardiac defect, namely truncus arteriosus with left aortic arch and hypoplasia of pulmonary arteries. Gupta et al. described a rare association of truncus arteriosus with VACTERL with longer duration of survival (5). Tracheoesophageal defects occur in approximately 50-80% of the patients (1). Nonetheless, the present case did not have this defect.

The type and severity of renal anomalies with VACTERL association vary widely. They are known to occur in approximately 50-80% of the patients.

The common anomalies include unilateral or

bilateral renal agenesis, horseshoe kidney, and cystic and/or dysplastic kidneys. Our case was suspected of ureteral and genitourinary anomalies. Few researchers stated that renal anomalies should not be considered as one of the defining components (1-4). Although the present case had structurally normal kidneys, he had renal failure for 10 days.

The limb malformations have been reported in approximately 40-50% of the patients. They include a displaced or hypoplastic thumb, polydactyly, syndactyly, and forearm defects (4). The most classic defects described are the radial anomalies (types I-IV). The severity of limb anomalies in the affected patients may vary widely. The present case had unilateral radial aplasia type IV. Carli et al. reported the incidence of complete radial aplasia in 33% of the patients (out of 25 cases) investigated in their series (4). Although many other malformations have been described in the affected patients, single umbilical artery may sometimes be the first antenatal clue to the diagnosis of VACTERL association.

The management of the patients with VACTERL association is a complex measure, which needs surgery in a staged manner. The treatment of this defect involves many issues related to each component feature (1). The components that would be incompatible with life, such as severe cardiac defects, imperforate anus, and tracheaesophageal defects, are managed with surgery in the immediate neonatal period. The defects like genitourinary anomalies are frequently treated in a staged manner (1). The correction of cardiac malformations may also require multiple surgeries, depending on the specific type of congenital defect.

The neonates with truncus arteriosus often present with congestive heart failure, as in the present case, requiring diuretics and digitalis. If truncus arteriosus is associated with the interruption of the aortic arch or aortic coarctation, they need prostaglandin infusion. Truncus arteriosus invariably requires operative repair. Nowadays, primary complete repair in the neonatal period is carried out in most of the advanced centers unlike palliative pulmonary artery banding in the neonatal period, followed by a complete repair in the infancy that is performed at older age.

Complete primary repair involves the closure of the VSD, committing the common arterial trunk to the left ventricle, and reconstruction of the right ventricular outflow tract. A survival rate of 90-95% has been reported in the North American centers after the complete repair of the truncus arteriosus (6). The cardiac surgery in the neonatal period is available only in selected centers in developing countries and is costly.

Even with the optimal surgical corrections of such malformations as cardiac anomalies, tracheoesophageal fistula, and limb abnormalities, the patients can face considerable medical challenges throughout life. However, component-specific sequelae usually do not give rise to neurocognitive impairment.

Conclusion

The components of VACTERL association may include new anomalies.

Acknowledgments

Nile.

Conflicts of interests

None.

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