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Case Report

Pentalogy of Cantrell: A Case Report of Probable Pentalogy of Cantrell in a Full-term Neonate

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ABSTRACT

Background: Pentalogy of Cantrell (PC) is an extremely rare congenital anomaly which was first described in 1985. The incidence of the PC has been reported to vary from 5.5-7.9 cases per million live births. This anomaly involves diaphragmatic pericardial defects, lower sternal defect, intracardiac anomalies, ventral abdominal wall abnormality, and anterior diaphragmatic defect. Considering the number of presented anomalies, PC is classified into three groups of certain, probable, or incomplete pentalogy. Although the etiology of PC is unknown, it has been assumed that abnormalities in intra embryonic mesodermal differentiation formation and migration at around 14-18 days after conception are responsible for the anomalies observed in PC. The prenatal diagnosis of the PC can be made via prenatal two-dimensional or three-dimensional ultrasound.

Case report: This case report investigated an Iranian 2-hour-old full-term neonate who was referred to Imam Reza Hospital, Kermanshah, Iran, due to respiratory distress and abdominal wall defect. The patient was finally diagnosed with a probable PC, although his prenatal investigations were considered normal.

Conclusion: Although ultrasonography is a widely available tool for prenatal diagnosis of PC, as in our case, the absence of ectopic heart and omphalocele makes the prenatal diagnosis of PC via ultrasonography less possible.

Keywords: Abdominal wall defects, Diaphragmatic defects, Pentalogy of cantrell

Introduction

Pentalogy of Cantrell (PC) was first described in 1985. The full spectrum of PC is characterized by five anomalies, namely 1)defects in the diaphragmatic pericardium with pericardio-peritoneal communication, 2) lower sternal defect, 3) various congenital intracardiac abnormalities, 4) ventral abdominal wall abnormality, and 5) anterior diaphragmatic defect (1). Pentalogy of Cantrell, considering the number of presented anomalies, is classified into three groups of certain, probable, or incomplete pentalogy. In this regard, if patients fulfill all five abnormalities, they are considered definite cases. However, patients with four defects, encompassing a ventral wall defect and an intra-cardiac abnormality, are defined as probable cases. Lastly, if patients lack either intra-cardiac defects or one or more of the explained abnormalities, they are determined as incomplete cases (2).

Case report

The patient was a 2-hour-old boy weighing 2,540 g, from a 21-year-old (gravida 2, para 2, abort 0) mother with the gestational age of 38 weeks, born via a cesarean section. The patient's parents were non-consanguineous marriage (28-year-old father). The mother had no special past medical history and adamantly denied any history of teratogen exposure, smoking, and alcohol or illicit drug use. Her first child was a 5-year-old healthy boy born via a C-section because of a breech presentation. She had undergone prenatal care and screening tests which were all normal. She did not need a fetal echocardiogram since her prenatal examinations were normal. No evidence of oligohydramnios or polyhydramnios incidence was detected. She mentioned no remarkable diseases in her family history. The neonate's APGAR scores at one and five minute were obtained as 4 and 7, respectively.

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The patient was referred to the neonatal intensive care unit because of respiratory distress and a suspicious abdominal wall defect. The examination revealed a full-term neonate with no obvious facial or skeletal dysmorphism and a supraumbilical 4×4 cm wall defect covered by intact skin (Figure 1). The examination of the heart and lungs revealed a III/VI degree systolic murmur and absence of the respiratory sounds on the right side lung. The subject's neurologic reflexes, including moro, grasp, and sucking reflexes, were reduced. The genitalia was in normal status and the anus was not imperforated and located in a normal anatomic region .

Chest radiographs showed a large opacity on the right side indicating a diaphragmatic hernia (Figure 2). Therefore, a computerized tomography scan was recommended for further evaluation of the chest, which was not possible due to the neonate's unstable status. The ultrasonography of the chest revealed the presence of the liver on the right side. Regardless of the upper section of the right hemithorax, the lung tissue was not detected. The heart was shifted to the left side. The diaphragmatic tissue was not completely discernible and defects were found in the anterior and mediastinal part. In the abdominal ultrasonography, moderate and mild hydronephrosis were found respectively in the right and left kidneys.

Figure 1. Photograph of the patient showing the supraumbilical defect

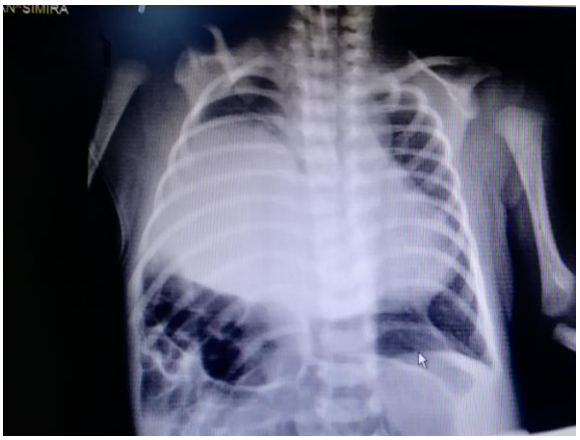

Figure 2. Chest radiograph showing a large opacity in the right side indicating a diaphragmatic hernia

The result of the cranial ultrasonography was normal. The findings of the echocardiography were as follows: a pericardial diaphragmatic defect, dilated right and left atrium, large perimembranous ventricular septal defect (VSD), large atrial septal defect (ASD), thin and atretic pulmonary artery (PA) with no outgoing blood flow, patent ductus arteriosus (PDA), dilated and large aortic artery, severe tricuspid valve regurgitation, and pulmonary hypertension.

The subject received antibiotics and milrinone for better cardiac output and was under mechanical ventilation. Due to his unstable condition, no operative procedures were conducted. The patient passed away on the third day of life as a result of a cardiopulmonary arrest. The patient's parents did not allow for an autopsy.

Discussion

Pentalogy of Cantrell is a rare congenital anomaly that was first described in 1985. The incidence of the PC varies from 5.5-7.9 per million live births (1-4). A history of the disease in the siblings have been reported in some cases with the provision of evidence for a familial inheritance (3, 4). According to Toyama, PC is classified into three classes, namely class I: presentation of all five defects, class II: presentation of four defects, including a ventral wall defect and an intracardiac abnormality, and class III: various combinations of the anomalies in the presence of sternal defect (2). In this study, the researchers presented a case of class II PC in a full-term neonate showing four of the anomalies, involving diaphragmatic pericardial defects, manifold intracardiac anomalies, ventral abdominal wall defect, and anterior diaphragmatic defect.

Cantrell described the common cardiac

anomalies from the lowest to highest prevalence as tetralogy of Fallot, left ventricular diverticulum, pulmonary atresia or stenosis, ASD, and VSD, respectively (1). Even though VSD is the most common cardiac anomaly in PC, it has been declared that this anomaly is not universally present .

Although the etiology of PC is unknown, it has been assumed that abnormalities in intraembryonic mesoderm differentiation formation and migration at around 14-18 days after conception account for the anomalies observed in PC (5). Despite the fact that most cases are observed sporadically, pieces of evidence of familial inheritance are available (6, 7). In our patient, neither a positive family history nor consanguineous marriage was present.

There has been an association between PC and chromosomal abnormalities, such as Turner syndrome, trisomy 18, and trisomy 21. Since the Wnt/β-Catenin pathway accounts for the migration of mesenchymal cells to the midline, the relationship between Goltz-Gorlin syndrome and PC can be attributed to the X-linked Drosophila porcupine homolog mutation, the gene that takes part in Wnt ligand acetylation (8-10). Additionally, the duplication of aldehyde dehydrogenase 1 family member A2 gene involved in vitamin A metabolism, which plays a critical role in diaphragm and heart development is identified in patients with PC. Some other associations, such as cystic hygroma, bilateral inguinal hernias, amniotic band syndrome, and exencephaly, have also been detected in patients with PC (5, 6).

The prenatal diagnosis of the PC can be made via prenatal two-dimensional or three-dimensional ultrasound (11, 12). The earliest time points for the prenatal PC diagnosis are reported to be the 9th and 11th gestational weeks (13, 14). However, in our case, all the prenatal ultrasounds were reported as normal. It is noteworthy the accurate diagnosis is highly dependent on the sonographer's expertise. Nevertheless, in this case, the absence of ectopic heart and omphalocele made the diagnosis of PC less possible in sonography. On the other hand, Gallot et al. reported the 54% sonographic prenatal detection rate of congenital diaphragmatic hernia (15). The prognosis and survival of these patients are very poor with a survival rate of hours rather than days and years (16, 17).

Conclusion

Although ultrasonography is a widely available tool for prenatal diagnosis of PC, as in our case, the absence of ectopic heart and omphalocele makes the prenatal diagnosis of PC via ultrasonography less possible.

Acknowledgments

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Conflicts of interest

The authors declare that there is no conflict of interest.

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