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8th Edition of International Conference on Clinical and Medical Case Reports - A rare case report of complete expression of pentalogy of cantrell: from radiology perspective

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Abstract

Pentalogy of cantrell consists of an extensive defect of the thoraco-abdominal wall, which has nearly always a lethal prognosis. The defect is characterized by the association of five anomalies: omphalocele, cardiac ectopia, absence of the distal portion of the sternum, absence of the anterior diaphragm and absence of the parietal diaphragmatic pericardium. It has a rare frequency of about 5.5 per 1,000,000 live births. There is a common association with intra cardiac anomalies such as ventricular septum defect, tetralogy of fallot and transposition of great vessels. The pathogenesis remains unclear. Here we present an imaging findings with antenatal two dimensional (2D) and three dimensional (3D) ultrasound and fetal magnetic resonance imaging (MRI) in a 20 weeks of gestation with a multiple anomalies, based on which the diagnosis of complete pentalogy of cantrell was given with a brief literature. Post mortem radiography, 3D computed tomography (CT) and clinical autopsy were performed additionally to enhance the visualization of fetal anomalies and to confirm the diagnosis. Extensive imaging of cardiac, thoracic and abdominal malformations by ultrasound and MRI is complementary for a clear diagnosis and counseling of the patient.

EMBRYOGENESIS

The sternum, abdominal wall, pericardium, and part of the diaphragm arise from somatic mesoderm, while the myocardium arises from splanchnic mesoderm. An event occurring prior to differentiation of the mesoderm into these two layers could produce defects in all of the involved structures, as seen in pentalogy of Cantrell. Although a specific etiology is unknown, the timing of the event or insult would be between 14 and 18 days after conception. The proposed embryogenesis postulates a failure of the lateral mesodermal folds to migrate to the midline, causing the sternal and abdominal defects, and failure of the septum transversum to develop, causing defects in the anterior diaphragm and pericardium.

DEFINITION:

The complete syndrome is characterized by two major defects: ectopia cordis and an abdominal wall defect (most commonly an omphalocele, but gastroschisis can also be present). The other three defects of the pentalogy are disruption of all the interposing structures: the distal sternum, anterior diaphragm and diaphragmatic pericardium. Incomplete expressions have also been reported.

Ultrasound diagnosis

Diagnosis of the complete syndrome requires the five criteria described by Cantrell, but incomplete variant forms exhibiting three or four of the features have been described. The sternal defect can range from absence of the xiphoid to cleaving, shortening, or absence of the entire sternum. The abdominal defect can range from a wide rectus muscle diastasis to a large omphalocele. The most common intracardiac defects are atrial septal defect, ventricular septal defect, and tetralogy of Fallot. The syndrome has been diagnosed prenatally , but as the defects range from subtle to severe, the ability to make the ultrasound diagnosis varies. Even at birth, the full extent of the syndrome may not be apparent, as the sternal defect may be minor and therefore without true ectopia cordis. In the case presented, a large open sternal defect with ectopia cordis was contiguous with the upper portion of the large gastroschisis.

Differential diagnosis

Differential diagnosis includes isolated ectopia cordis, isolated abdominal wall defect, amniotic band syndrome, and body stalk anomaly. The syndrome should be considered with any diagnosis of omphalocele or ectopia cordis. The key features for distinguishing these conditions is the position of abdominal wall defect in relation to the umbilical cord, eviscerated organs, the presence or absence of membranes or bands, and associated anomalies. Omphalocele usually involves a midline defect at the umbilical cord insertion with a covering membrane. Gas-

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troschisis is a midline or lateral abdominal wall defect with cord inserted just below the defect. An eccentric large lateral defect and adherence of the placenta to the defect is typically present in body stalk anomalies. The presence of an unexplained ventral wall defect along with extremity deformity with an adherent band suggests amniotic band syndrome.

Associated anomalies

If a diagnosis is made by ultrasound, chromosomal analysis is recommended. Associations with trisomy 18, trisomy 13, and Turner syndrome have been reported. Careful imaging should be performed to rule out associated anomalies. Fetal echocardiography is indicated to evaluate the extent of any intracardiac abnormalities8. Others include cranial and facial anomalies, clubfeet, malrotation of the colon, hydrocephalus, and anencephaly.

Management

In view of the poor prognosis, termination of pregnancy can be considered if ultrasound diagnosis is made before viability. In patients choosing to continue the pregnancy, there is no data indicating improved or changed outcome with cesarean delivery.

After delivery, repair of the gastroschisis and omphalocele should not be delayed. Repair of the sternal, diaphragmatic, and pericardial defects can be attempted at the same time. Surgical correction is often difficult secondary to hypoplasia of the thoracic cage and inability to enclose the ectopic heart. Some affected infants have respiratory insufficiency secondary to pulmonary hypoplasia. Recognition and treatment of any intracardiac anomaly is important, as congenital heart disease is a source of major morbidity in infants surviving the neonatal period.

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