Ultrasound Meets Magnetic Resonance Imaging in the Diagnosis of Pentalogy of Cantrell with Complete Ectopy of the Heart

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Abstract

We report a case of a complete pentalogy of Cantrell at 25 weeks of gestation through antenatal Two Dimensional (2D) and Three Dimensional (3D) ultrasound and Fetal Magnetic Resonance Imaging (MRI). A complete ectopiacardios with a supraumbilical hepato-omphalocele were diagnosed. Echocardiography showed multiples cardiac anomalies: a large ventricular septum defect, a hypoplastic right ventricle, a transposition of the great vessels with a small right outflow.

Prenatal and post mortem MRI and 3D Computed Tomography (CT) were performed in addition to routine 2D ultrasound to enhance the visualization of fetal anomalies for accurate diagnosis.

Fetal autopsy following termination of the pregnancy confirmed the presence of all malformations detected prenatally.

In our opinion extensive imaging of cardiac, thoracic and abdominal malformations by ultrasound and MRI is complementary for a clear diagnosis and counseling of the patient.

Keywords: Pentalogy of cantrell; Two dimensional ultrasound; Three dimensional ultrasound; MRI

Case Report

A 31-year-old woman, gravida 3 para 2, was referred to our center because of a suspicion of a fetal anomaly at 25 weeks of gestation. Up to this point the pregnancy course was unremarkable. Previous medical and obstetric histories were unremarkable except for 2 caesarian deliveries a term. Detailed two and three dimensional transabdominal ultrasound revealed a live female fetus with measurements consistent with 25 weeks of gestation. Amniotic fluid index and placenta were normal. Fetal heart was completely displaced outside the thoracic cavity with the cardiac apex pointing toward the fetal chin. Fetal heart examination showed a hypoplastic right ventricle, a ventricular septum defect, a transposition of the great arteries with a hypoplastic right outflow (Figure 1a). A supraumbilical omphalocele with partial excision of the liver was also diagnosed (Figure 1b). A three dimensional reconstruction showed the heart protruding through the sternum and confirmed the omphalocele (Figure 1c).

An amniocentesis was performed for karyotyping and molecular genetic analysis.

Fetal MRI reported a normal central nervous system without arguments for spinal defects. MRI confirmed the ectopic position of the heart and the presence of an important lower sternal and abdominal wall defect. The sternal manubrium was present, with only a few zones of ossification at the corpus of the sternum. There was no visualization of the distal part of the sternum. From the dynamic TrueFisp imaging a hypoplastic right ventricle with associated parallel position of the great arteries of the cardiac outflow was seen. Secondary to the ectopic heart we found a tiny thoracic cavity with normal lungparenchyma. Pericardium was found to be intact, and there was no suspicion for a diaphragmatic defect on MRI (Figure 2).

Karyotyping on amniotic cells was performed by G-banding and was found normal. Molecular karyotyping by array-Comparative Genome Hybridization (CGH) showed no abnormalities.

After discussion of the findings and multidisciplinary counseling about the poor prognosis of this case, the patient opted for a termination of pregnancy.

Post mortem MRI and 3 D CT reconstruction confirmed the ectopiacardios with associated sternal defect and the omphalocele with partial herniation of the liver (Figure 3). Fetus and placenta underwent postmortem examination. The fetus weighed 1030 g and was 360 mm long. External exploration showed a large thoraco-abdominal wall defect that involved the inferior sternal portion and the abdominal wall to the supraumbilical zone. The infant’s heart was confirmed to protrude completely through a defect

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Discussion

The pentalogy of Cantrell is a rare and severe congenital disorder. This congenital disorder was first described in 1958 by Cantrell and it is characterized by the presence of two major defects: ectopia cordis and an abdominal wall defect [1]. The syndrome has an estimated incidence of 5.5 per 1 million live births and only a few patients with a full spectrum of the pentalogy have been described [1,2]. The condition consist a combination of five congenital anomalies: a midline supraumbilical abdominal wall defect, a defect of the lower sternum, a deficiency of the anterior diaphragm and a congenital heart anomaly [1].

The etiology and pathogenesis of the pentalogy of Cantrell is still not fully known. The cause of the defect seems to be a developmental failure of the ventral thoraco-abdominal wall closure in the primordial lateral mesodermal tissue during embryological stage [1]. A rupture of the chorion or yolk sac is also suggested to prevent proper midline fusion of the chest wall [3].

Although sporadic in most of the described infants, chromosomal abnormalities have also been reported. X-linked recessive inheritance was suggested for some recurrent cases and some genes located on the X-chromosome (Xq25-q26.1) may be involved [4].

Different intracardiac anomalies are associated (i.e. tetralogy of Fallot, ventricular septum defect, atrial septum defect, ventricular diverticle). Other craniofacial and central nervous system anomalies have been reported such as cleft lip and/or palate, cystic hygroma, encephalocele, hydrocephalus, cranioschizis and anencephaly. Limb defects, clubfeet and abdominal organ defects such as malrotation of the colon and dysplastic kidney have also been described [2].

The prognosis is dependent on the severity of the abdominal wall defect, the intracardiac malformations as well as the presence of associated anomalies. Minor degrees of ectopia cordis may be surgically correctable. Prognosis of cases presenting a complete extrusion of the heart and abdominal contents are extremely poor [5].

In this case we found that 2D,3D ultrasound and fetal echocardiography were valuable in making the diagnosis as well in counseling the patient. The 3D reconstructed imaging did provide a complete view of the anomaly. This prenatal visualization allows the patient to better understand the diagnosis. During pregnancy some measurement are very difficult by ultrasound because of the fetal position, the maternal constitution and the size of the defect. While MRI and CT can provide a better field of view.

In this case MRI offers an optimal assessment of this syndrome confirming and defining ultrasound findings and revealed supplementary diagnostic information with regard to the absent lower part of the sternum, the pericardium and the diaphragm. The reconstructed imaging did provide a comprehensive view of the anomaly which was useful in parental counseling.

3D CT reconstruction, prenatal and postmortem MRI are important teaching tools, especially in an academic setting such as our center. They provide important structural information, measurements and visualization of critical anatomic details. In our opinion extensive three dimensional imaging of cardiac, thoracic and abdominal malformations by ultrasound and MRI is complementary for a clear diagnosis of the

Figure 2: Fetal MRI using single shot turbo spin echo, shows the heart in an ectopic position (*) and eversion of a part of the liver at the midline (arrow). Notice the small thoracic cavity (t). No malformation of the central nervous system could be seen.

Figure 3: Postnatal MRI (a), 3D CT (b) and classical autopsy (c) confirmed the transposition of the great arteries (*) and the extravisceration of heart and liver.

Figure 4: Atrial septum defect (I), ventricular septum defect (II), overriding aorta (III) and transposition of the great vessels (IV) (RV: right ventricle, LV: left ventricle, RA: right atrium, LA: left atrium, AO: aortic outflow tract, PO: pulmonary outflow tract)
extent of the defect in order to define possible preoperative planning and counseling the patient. Detailed post mortem examination and autopsy are important for final confirmation of the diagnosis and for academic teaching [6-8].

References


