Pentalogy of Cantrell Associated With Thoracoabdominal Ectopia Cordis

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A 4-day-old female infant with a partially extrathoracic heart in her supraumbilical abdominal wall (Figure 1A) presented with symptoms of tachypnea and mild cyanosis. Physical examination showed a thoracoabdominal wall defect with thoracoabdominal ectopia cordis. The xyphoid was absent and the sternum was short. Oxygen room air saturation was 85%. Chest x-ray showed dextrocardia and descensus of the heart contour to the right epigastrium (Figure 1B). Helical computed tomography (CT) demonstrated a ventral thoracoabdominal wall defect with the 2 ventricles protruding through the defective anterior diaphragm to epigastrium and positioned on the liver (Figure 1C through 1E). Truncus arteriosus and persistent left superior vena cava were observed on helical CT imaging (Figure 1C and 1F). Also present were ventricular septal defect, atrial septal defect, patent ductus arteriosus, and overriding aorta. Magnetic resonance imaging and echocardiography also successfully visualized truncus arteriosus (Figure 1G and 1H; also, see Figure 2 and online-only Data Supplement Movies I through III). Repeated echocardiogram confirmed these intracardiac findings and revealed the absence of the anterior part of both pericardium and diaphragm. Chromosome study showed a normal female karyotype.

The child was stabilized with prostaglandin E1 infusion with an acceptable oxygenation. The primary decision was made to apply a strict follow-up and perform elective palliative operation at the age of 3 to 6 months, but her parents signed a “do not resuscitate” order in view of the poor prognosis and the patient died on day 61 from cardiopulmonary failure.

Pentalogy of Cantrell is a congenital malformation characterized by (1) lower sternum defect, (2) anterior diaphragm defect, (3) parietal pericardium defect, (4) omphalocele, and (5) congenital heart anomalies. Ectopia cordis might or might not be associated with pentalogy of Cantrell. This pentalogy occurs with various degrees of severity, ranging from incomplete to severe expression with involvement of other organ systems. Most cases are sporadic. The exact pathogenesis remains unknown to date. No familial tendency has been demonstrated. This case is a full spectrum of the pentalogy of Cantrell.

Prognosis depends on the severity of the cardiac and associated malformations. Cardiac anomalies can vary from a single intracardiac defect to the most severe malformation of complete ectopia cordis in combination with complex intracardiac defect. In patients with complex cardiac malformations who undergo surgery during the first days of life, mortality can be as high as 50%. Cardiac rupture, tamponade, sudden death, endocarditis, peripheral embolism, heart failure, and arrhythmia have all been described as complications and causes of death. Our case presented with thoracoabdominal ectopia cordis in association with truncus arteriosus, ventricular septal defect, atrial septal defect, patent ductus arteriosus, and persistent left superior vena cava. This complex combination made the prognosis much worse.

Diagnosis of pentalogy of Cantrell is possible on antenatal sonography in the first trimester of pregnancy. Accurate early anatomic assessment of components of this syndrome is crucial for optimal parental counseling and decision making on the outcome of pregnancy. Antenatal ultrasound, echocardiography, and magnetic resonance imaging when necessary should be applied to search for this combination of malformations.

The surgical intervention consists of corrective or palliative cardiovascular surgery, correction of ventral hernia and diaphragmatic defects, and correction of associated anomalies. Generally, pentalogy of Cantrell is fatal without surgery. Successful surgery has been accomplished despite its high mortality rate, but the surviving cases did not have, or had only minor degrees of, ectopia cordis with small omphaloceles and simple intracardiac anomalies. Nevertheless, surgical intervention is the treatment of choice to prevent potentially catastrophic complications. The best treatment strategy depends on the size of the...
Figure 1. A, Image of the extrathoracic heart on day 12. B, Contour of heart was not in the left thoracic cavity but situated in the right epigastrium. C, Helical CT demonstrated evisceration of the 2 ventricles out of thorax and superior to the liver. Truncus arteriosus was clearly visualized. D, E, and F, Helical CT 3-dimensional imaging of the ectopic heart showed the 2 ectopic ventricles from views of left anterior, right posterior, and anterior, respectively. Also present were aorta, inferior vena cava, and hepatic veins. Right superior vena cava and persistent left superior vena cava were also visible. G and H, Truncus arteriosus was demonstrated by magnetic resonance imaging and echocardiography, respectively. TA indicates truncus arteriosus; RV, right ventricle; LV, left ventricle; DA, descending aorta; IVC, inferior vena cava; RSVC, right superior vena cava; LSVC, left superior vena cava; RA, right atrium; and LA, left atrium.
abdominal wall defect, the associated heart anomalies, and the type of ectopia cordis.

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Disclosures
None.

References
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