Assessment of Pentalogy of Cantrell using 3D Multidetector Computed Tomography

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Abbreviations: MDCT, multidetector computed tomography

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Abstract

A 7-month-old white female with Pentalogy of Cantrell was imaged using 64 slice multidetector computed tomography (MDCT) with 3D mapping to better determine the extent of cardiac, thoracic, and abdominal malformations. Complimentary to ultrasound, the use of 3D 64 slice MDCT can facilitate effective diagnosis and treatment planning in cases of Pentalogy of Cantrell.

Introduction

Pentalogy of Cantrell, also known as thoracoabdominal or Cantrell-Haller-Ravich Syndrome, is a spectrum of midline thoracoabdominal defects. These congenital malformations consist of defects in the supraumbilical wall, lower sternum, anterior diaphragm, and diaphragmatic pericardium, often resulting in parietal herniation of the heart accompanied by other cardiothoracic abnormalities [1, 2, 3]. Ventricular septal defects, atrial septal defects and, on occasion, tetralogy of Fallot are associated with these malformations. Craniofacial and central nervous system malformations, such as anencephaly and craniorachischisis, along with a wide range of sporadic abnormalities, including intestinal atresia and renal agenesis, are also found in this syndrome [1, 2, 4]. Pentalogy of Cantrell often occurs with an omphalocele of the anterior abdominal wall superior to the umbilicus, likely attributed to failure in the proper development of mesoderm around day 14-18 post-gestation [1, 5]. Survival is contingent on the number and severity of structural anomalies, especially those intrinsic to the heart, and this spectrum of defects is divided into complete or partial pentalogy of Cantrell. Complete pentalogy classically presents with two major defects, severe ectopia cordis with intracardiac abnormalities and abdominal wall defects, accompanied by defects disrupting the

sternum, diaphragm and pericardium [6, 7, 8]. All other variations are considered partial or incomplete and cardiac anomalies are usually absent. We present the utility of the volumetric patient examination using 3D 64 slice MDCT in order to determine the optimal patient care in a rare case of incomplete pentalogy of Cantrell.

Case Report

An 18-year-old female in foster care whose past medical history was unremarkable except for asthma and a previous abortion was referred to our institution during her second trimester of pregnancy. A fetal ultrasound suggested pentalogy of Cantrell. This sonogram showed a female fetus with a large upper anterior abdominal wall defect (omphalocele) containing liver and stomach with marked ectopia cordis (Figure 1). The mother and fetus were closely monitored for cardiac, pulmonary, and gastrointestinal complications until a primary low segment transverse cesarean section was performed at 40 weeks gestation.

A vigorous white female infant was born, weighing 2,650 grams with Apgar scores of 7 and 8, and presenting with incomplete pentalogy of Cantrell. Based on sonograms, radiographs, and a voiding cystourethrogram, this case was characterized by a structurally normal heart with partial ectopia cordis, omphalocele, sternal dehiscence, anterior diaphragmatic hernia, gastroesophageal reflux, grade 2 vesicoureteral reflux, hydrocephalus, and chronic lung disease with ventilator dependence. The diagnosis of incomplete or partial pentalogy of Cantrell was made based on the absence of significant congenital intracardiac disease. A mildly abnormal pulmonary valve and moderate sub-valvar pulmonary stenosis was observed on ultrasound with no noticeable ventricular septal defects. On post partum day 1, a cross table lateral radiograph was taken before the omphalocele was repaired and the heart enclosed in surrounding fascia and subcutaneous tissue (Figure 2). The sternal defect inferior to the heart was not repaired and a prosthesis was placed over the chest wall for protection. The infant was monitored for the next 18 months, during which she was hospitalized for worsening respiratory distress marked by hypoventilation and cyanosis intensified by various infections and secondary complications. 3D multidetector computed tomography (dual-source 64 slice MDCT, Siemens Medical Systems®) was performed at 7 months to assess the cardiac, lung and abdominal abnormalities using 64 slice with 3D post processing (Figures 3, 4, 5). Cyanosis was suspected to be the result of pulmonary hypoplasia with secondary chronic lung disease and pulmonary hypertension. Based on the MDCT findings, her hypoxia was further exacerbated by the presence of persistent moderate subvalvar right ventricular outflow tract stenosis. This was localized to the region where the right ventricular outflow tract crossed posterior to the closed sternum (Figure 5). Pulmonary hypertension was diagnosed likely due to the patient's chronic lung disease. Her complex condition was treated conservatively with a tracheostomy for mechanical ventilation to support acute and chronic respiratory failure.

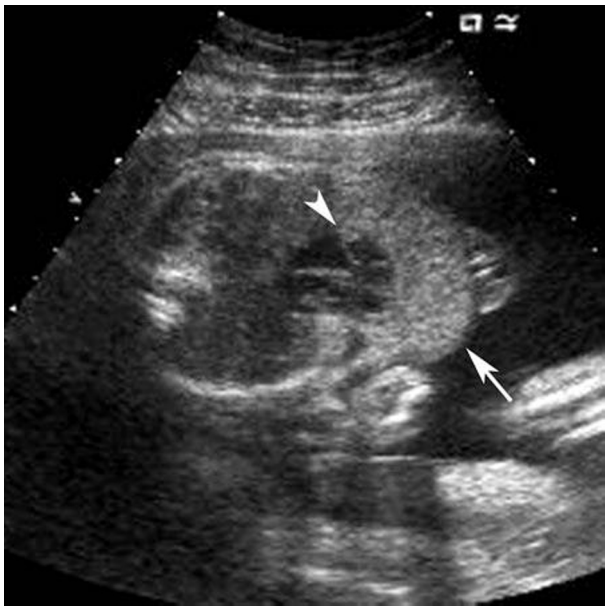

Figure 1. Female with Pentology of Cantrell. Axial prenatal sonogram image of the fetus at week 33 with ectopia cordis (arrowhead) and omphalocele (arrow) containing liver.

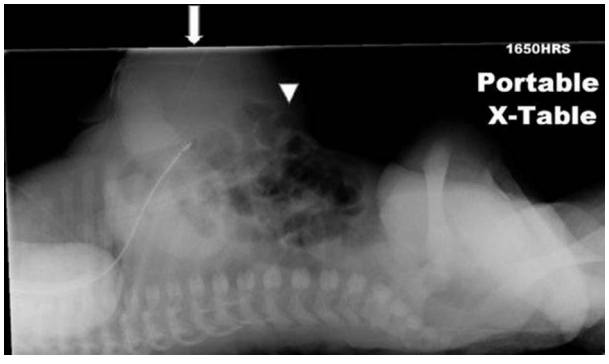

Figure 2. Female with Pentology of Cantrell. Cross-table lateral radiograph of the thorax and abdomen of the infant shortly after birth. The ectopia cordis (arrow) on the left is severe with an adjacent smaller extruding gastrointestinal omphalocele (arrowhead).

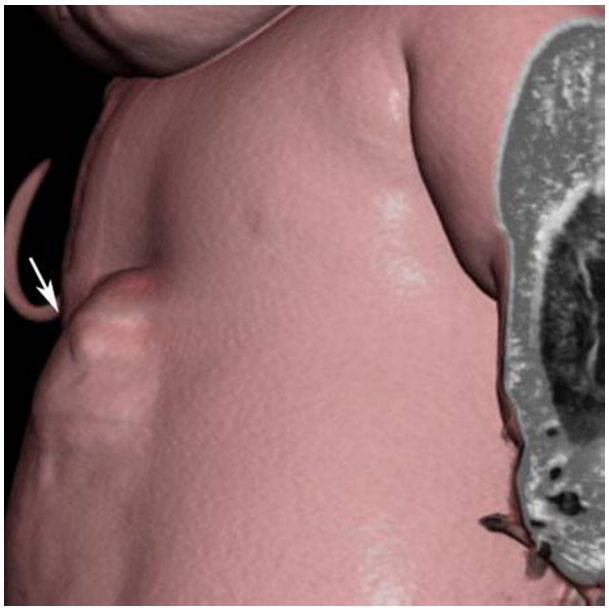

Figure 3. Female with Pentology of Cantrell. 64-slice volume-rendered MDCT image of the patient 7 months postpartum, showing the external position of the anterior heart beneath the skin (arrow).

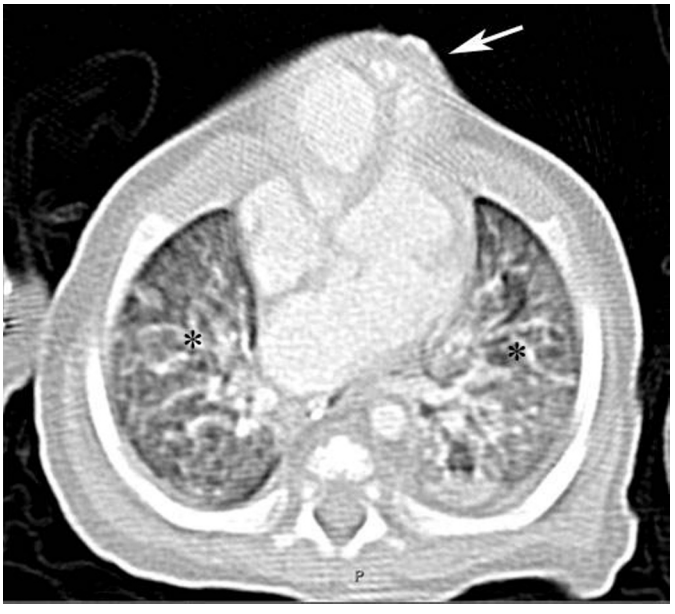

Figure 4. Female with Pentology of Cantrell. Thoracic cross-section of the patient 7 months postpartum, showing the ectopia cordis (arrow) external to the rib cage and diffuse chronic lung disease due to pulmonary hypoplasia (asterisks).

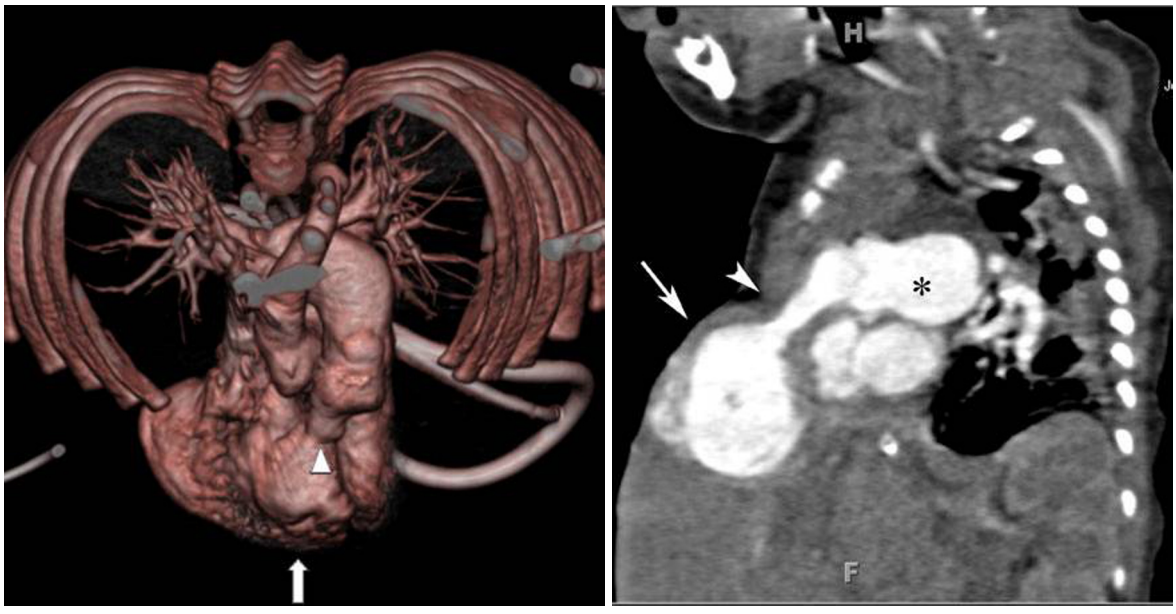

Figure 5. Female with Pentology of Cantrell. 64 slice volume rendered (a) and reconstructed maximum intensity projection (MIP, b) MDCT images of the patient 7 months after birth showing ectopia cordis (arrow) with CT angiography. The external position of the heart and subpulmonary stenosis caused by the indenting sternum (arrowheads) are notable with post-stenotic main pulmonary artery dilatation (asterisk).

Discussion

A rare yet noteworthy syndrome, pentalogy of Cantrell can be associated with aneuploidy, such as trisomy 18 and other X-linked defects that have not been extensively characterized due to the rare nature of this condition [9, 10]. Prognosis is dependent on the number and severity of defects, especially abnormalities associated with the heart, and can be categorized as complete or partial pentalogy of Cantrell. Complete pentalogy requires the presence of two major defects, severe ectopia cordis and abdominal wall defects, usually an omphalocele or gastrochisis, and three other defects causing disruption of the sternum, diaphragm and diaphragmatic pericardium [6, 7, 9]. All other cases with milder defects are considered incomplete. Pentalogy of Cantrell is only seen in about 1 in 5.5 million live births and survival is largely dependent on the severity of the cardiac defect [5]. Prenatal diagnosis of this entity is currently based on prenatal ultrasound imaging, although diagnosis may be difficult depending on the severity of the anatomic defects. Magnetic resonance imaging in conjunction with echocardiography has been explored as a useful supplemental imaging modality to delineate cardiac and thoracic involvement [9]. MDCT provides a fast comprehensive 3D virtual exam of the anatomical complications associated with pentalogy of Cantrell. It should only be used complimentary to ultrasound and MRI, however, because of the relatively high radiation exposure in these infants [11]. On the other hand sedation is frequently required in infants undergoing MRI, which is usually not necessary with the current generation of CT scanners with fast image acquisition speed. This is a significant advantage in cases of cyanosis where sedation may lead to further respiratory depression.

Our case has partial ectopia cordis and a successfully repaired omphalocele without severe congenital heart disease, which may explain her ability to survive. Survival is poor (8-37%) in patients with complete pentalogy, characterized by severe ectopia cordis and abdominal wall defects associated with disruption of the distal sternum, anterior diaphragm, and diaphragmatic pericardium. The

prognosis is better for incomplete forms as in our case (60% survival) [6, 7, 8]. Due to the poor prognosis of Cantrell, termination of pregnancy remains an option for parents after an ultrasound diagnosis is made [8]. Omphalocele repair should not be delayed due to a risk of infection but is often difficult to attempt soon after birth since infants usually present with a hypoplastic thoracic cage unable to enclose the lungs and ectopic heart [8]. This particular case was complicated by a moderate subvalvar right ventricular outflow tract stenosis as well as chronic lung disease.

3D 64 slice MDCT was particularly useful in the assessment of the pulmonary artery outflow tract stenosis caused by the anterior thorax wall. This narrowing likely contributed to pulmonary hypertension and cyanosis together with the patient's pulmonary hypoplasia. 3D MDCT mapping may assist individualize therapy by providing useful anatomical information for optimizing patient care.

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