Pentalogy of Cantrell with Ectopia Cordis: CT Findings

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ABSTRACT

A 14-month-old girl with pentalogy of Cantrell, a very rare congenital syndrome characterized by an epigastric omphalocele and malformations of the heart, sternum, pericardium, and diaphragm, underwent echocardiography and multidetector computed tomography before surgical repair of these deformities was attempted. These tests revealed multiple cardiovascular and noncardiovascular abnormalities. After surgery, the patient's cardiovascular status was stable. Although studies have shown that echocardiography, multidetector computed tomography, and magnetic resonance imaging may each play a role in the diagnosis and management of this condition, there are few data available to support the use of one imaging modality over another.

CASE REPORT

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A full-term female infant presented with a complex ventral wall hernia defect after Cesarean delivery. Although prenatal ultrasound during the 19th week of gestation had revealed no irregularities, echocardiogram performed after delivery demonstrated multiple abnormalities including mesocardia, aneurysmal atrial septal tissue, elongated left-sided apex extending into the abdominal wall defect, double outlet right ventricle (DORV) with large conoventricular ventricular septal defect with inlet extension, pulmonary annulus and main pulmonary artery hypoplasia. Functional evaluation revealed a peak pressure gradient of 70 mmHg across the right ventricular outflow tract (normal <10mmHg). Additional vascular abnormalities found on echocardiogram included reverse-angle; long, tortuous patent ductus arteriosus and left aortic arch with an aberrant right subclavian artery. A follow-up multisequence, multiplanar MRI with intravenous gadolinium of the thorax and abdomen was immediately performed; this study demonstrated dextropositioning of the heart, with the root of the ascending aorta projecting over the right ventricle. Post contrast MRI revealed segments of myocardial atresia contributed from the anterior aspects of the right and left ventricles through presumed diaphragmatic and pericardial defects (later confirmed by surgical findings). MR images of the upper abdomen demonstrated portions of the left hepatic lobe and nonobstructed small bowel loops herniating anteriorly through an abdominal wall defect, with very little (if any) subcutaneous tissue covering these structures. The hepatic, splenic, and renal parenchyma demonstrated normal signal characteristics. A second echocardiogram, performed when the patient was 25 days of age, demonstrated D-malposition of the great vessels, with the aorta located anterior and to the right of the pulmonary artery; absence of the patent ductus arteriosus was also reported.

The patient remained stable and was treated as an outpatient for 5 months, presenting with occasional cyanotic

spells that gradually increased in frequency and severity. At 7 months of age, a pulmonary balloon angioplasty was performed, but symptomatic improvement was not seen. Surgical repair of the DORV, including ventricular septal defect closure to the right-sided aorta, placement of right ventricle to pulmonary artery homograft, left pulmonary arterioplasty, closure of the two 5-mm atrial septal defects, and ligation of the left superior vena cava, was performed. The patient's postoperative course was complicated by left-sided cerebral ischemic injury. Additional surgical management of noncardiac malformations was deferred. Episodes of oxygen desaturation recurred at 14 months of age; when echocardiography demonstrated severe homograft stenosis and moderate tricuspid regurgitation. A non-cardiac-gated, contrast-enhanced multidetector CT (MDCT) of the chest and abdomen was performed, revealing a severe narrowing of the conduit extending from the right ventricle to the right pulmonary artery. In addition, the MDCT study showed cardiomegaly with a massively dilated right atrium, a hypoplastic right ventricle, a hypoplastic left pulmonary artery, and a left-sided aortic arch arising from the left ventricle with an aberrant right subclavian artery. Diverticulae were also identified extending inferiorly from the heart, one in the midline extending inferiorly from the hypoplastic right ventricle and one just to the left of midline extending inferiorly from the left ventricle. The diverticulae arose inferior to the level of the xiphoid process, extending approximately 8.3 cm from the inferior margin of the xiphoid process and down the midline abdomen just below the skin surface into the omphalocele, terminating at the L4-L5 disk space level. This CT also revealed a covered midline abdominal wall defect consistent with an omphalocele, which contained part of the left lobe of the liver along with a portion of the small bowel and colon. The diverticula were noted anteriorly within the herniated cavity, with their tip terminating just below the inferior margin of the left lobe of the liver, which resided within the omphalocele. Other than the left gastric artery arising directly from the abdominal aorta, the vasculature appeared to be normal.

After this CT examination, the patient underwent revision of the right ventricular to pulmonary artery conduit, a pericardial-patch left pulmonary artery arterioplasty, closure of two small residual atrial septal defects, dissection and transection of the right and left ventricular diverticula, primary repair of the central diaphragmatic hernia, and prosthetic patch closure of the omphalocele. Currently, the patient's cardiovascular status is stable, and she is progressing well. A second stage of omphalocele closure is the next planned step of surgical management.

DISCUSSION

Pentalogy of Cantrell is a rare syndrome that was first described in 1958 and is classically characterized by 5 congenital anomalies: intracardiac defect, sternal cleft, pericardial defect, diaphragmatic defect, and epigastric omphalocele[1,2].

Etiology & Demographics

The pentalogy of Cantrell (also referred to as Cantrell's pentalogy or the Cantrell syndrome) has an estimated incidence of 1 per 65,000 live births [3]. In general, the complex nature of this disease makes it fatal unless there is prompt surgical intervention. The prognosis of each case is related to the severity of the diaphragmatic, pericardial, and intracardiac defects that characterize this condition. A developmental failure of the segmental mesoderm at approximately 14 to 18 days after conception (the time period for differentiation of somatic and splanchnic mesoderm) is believed to be responsible for these defects. Failure of the transverse septum (arising from the mesoderm) to partially or entirely complete the process of flexion or ventral folding is believed to cause the ventral diaphragmatic defects. Abdominal wall and sternal defects can also occur as a result of disrupted mesoderm development involving failure of ventral migration [1,2].

Clinical & Imaging findings

Not all cases of pentalogy of Cantrell present with these 5 classical findings [1,2]. In 1972, Toyama classified Cantrell's pentalogy into 3 different groups based on the expression of symptoms: class I presents with all 5 defects and is a definite diagnosis; class II presents with 4 of the 5 defects and is a probable diagnosis; and class III presents with varying combinations of defects and is considered an incomplete expression. (Table 2) Lacking the sternal defect, the patient in this report represents a case of class II pentalogy of Cantrell [4].

In utero and neonatal imaging studies play a key role in not only prenatal counseling, but also in assisting surgeons to formulate better operative plans for patients with this condition. Two-dimensional (2D) ultrasound is the imaging tool most often used to identify the characteristic malformations associated with pentalogy of Cantrell. In a 12-case study, Bonilla et al. concluded that, although the original diagnosis of pentalogy of Cantrell was accurate based on 2D ultrasonography in 11 of the patients, additional information was obtained by 3-dimensional (3D) ultrasonography in all cases; the investigators suggested that such additional information could be useful for prenatal counseling and postnatal therapeutic planning [5]. More recently, Desselle et al. reported a case in which correlation of 2D and 3D sonographic findings helped with successful neonatal surgical management [3,6]. These authors suggested that a second- and third-trimester 3D sonogram may be useful not only in assessing the volume and content of the omphalocele, but also in evaluating the size of the parietal defect, giving the surgeons an indication for primary or staged repair. In our patient, second-trimester in utero ultrasound examination failed to identify cardiac or vascular abnormalities.

Other imaging modalities have also been studied for the evaluation of pentalogy of Cantrell. McMahon et al. reported a case in which fetal magnetic resonance (MR) plus prenatal echocardiography allowed for optimal assessment of a fetus with ectopia cordis, providing prognostic information and improved preoperative planning. [7] As an alternative to MR,

MDCT may also be used in patients with pentalogy of Cantrell. Santiago-Herrera et al. used MDCT to evaluate the complex cardiac anatomy of a 17-month-old with pentalogy of Cantrell complicated with tetralogy of Fallot, pointing out that MDCT generally provides high spatial resolution, truly multiplanar capabilities, and fast acquisition time without the need for anesthesia. [8] However, the ionizing radiation and iodine contrast required for MDCT must be weighed against these benefits. In our patient, MRI confirmed the cardiac findings seen on echocardiogram and identified additional anatomic, vascular, and functional findings, which aided in assessment and surgical management of the condition; MDCT provided information about post-surgical status and the progression of abnormalities.

This case was complicated by several cardiac abnormalities, a few of which are among the rarest types reported. Assessing the anatomy within the abdomen and thorax with various imaging modalities provided valuable details to determine the type of repair needed for abdominal defects. It remains unclear whether CT scanning is more or less appropriate than MRI in the evaluation of patients with pentalogy of Cantrell. Given the rare nature of this condition, data are not available to support one technique over the other. The multidisciplinary team caring for the patient should therefore make such decisions based on risks of anesthesia (more likely needed for MR than CT), radiation dose, use of iodine contrast, and the type and level of anatomic detail needed for preoperative planning.

Treatment & Prognosis

Corrective or palliative surgery of the arrested development is vital, however the timing of surgery is determined by the severity of presentation. If delay of operative repair is possible, it is advisable to wait until 2 years of age. This delay allows for some growth of the thoracic cavity and avoidance of high intrathoracic pressures during repositioning of the heart. [9]

Surgical management consists of repair of the cardiac anomalies, the sternal cleft, and the diaphragmatic defect, Sometimes; one-stage surgical repair of all malformations is possible.

The reconstruction of intracardiac defect is a complicated surgery that requires cardiopulmonary bypass, deep hypothermia, and cardiopulmonary arrest. However, the resection of the ventricular diverticulum is crucial to prevent spontaneous rupture and sudden death by tachyarrhythmia. [10]

To reconstruct the sternal cleft, grafts or flaps harvested from the anterior thoracic wall are recommended due to a lower risk of infection. [11] Direct closure of diaphragmatic defect can be performed.

The prognosis of patients with pentalogy of Cantrell is dependent on the severity of the intracardiac abnormalities and associated malformations. Complex cardiac malformations have mortality as high as 50% during first days of life. [4]

Differential Diagnoses

The thoraco-abdominal wall results from the fusion of five distinct body folds at the base of the umbilical cord. Failure of fusion of the cephalic fold results in ectopia cordis, the complete or partial displacement of the heart outside the thoracic cavity. Failure of further growth may include ectopia cordis and the other four defects of pentalogy of Cantrell, or one of it variants. [9,12]

Other syndromes involving the anterior body wall such as 'omphalocele, exstrophy, imperforate anus, spinal defects' (OEIS complex) and limb-body wall complex, don't compromise the cardiac structures. These features aid the diagnosis. (Table 3)

TEACHING POINT

Pentalogy of Cantrell is a rare syndrome characterized by intracardiac and thoracoabdominal wall malformations. The prognosis as well as success of surgical intervention of these midline developmental defects depends on their severity. MDCT can provide multiplanar capabilities and fast acquisition time, to determine with accuracy the anatomy and vascularization.

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FIGURES

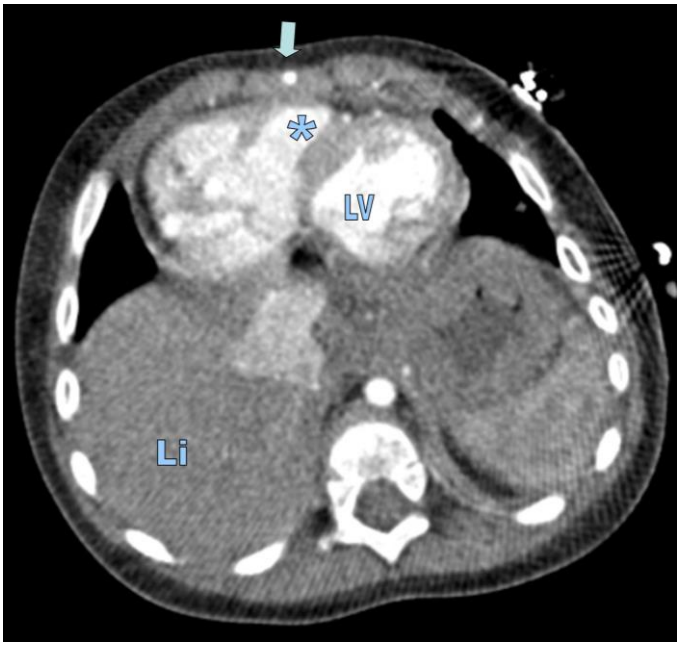


Figure 1: 14-month-old girl with Pentalogy of Cantrell. Axial image at the level of xiphoid process (arrow) and hepatic dome (Li) shows elongated left- and right-sided apex (*) extending anterior to the right ventricle. (Technique: Siemens Sensation 64; slice thickness 1.5mm, KvP 120; mAs 80-100; contrast Isovue 370, iopamidol.).

Figure 3 (right): 14-month-old girl with Pentalogy of Cantrell. Oblique Maximum Intensity Projection (MIP) image of the heart shows severe narrowing of the right ventricle-pulmonary artery conduit (arrow). (Technique: Siemens Sensation 64; slice thickness 1.5mm, KvP 120; mAs 80-100; contrast Isovue 370, iopamidol.).

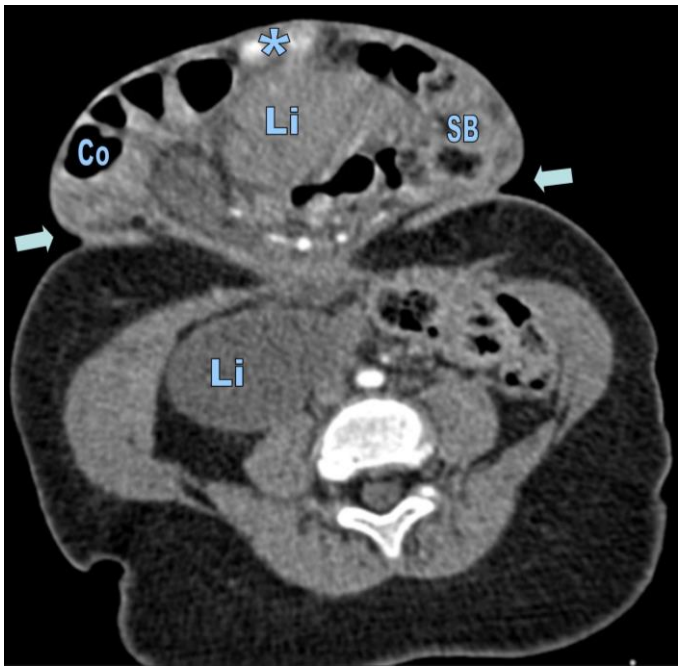
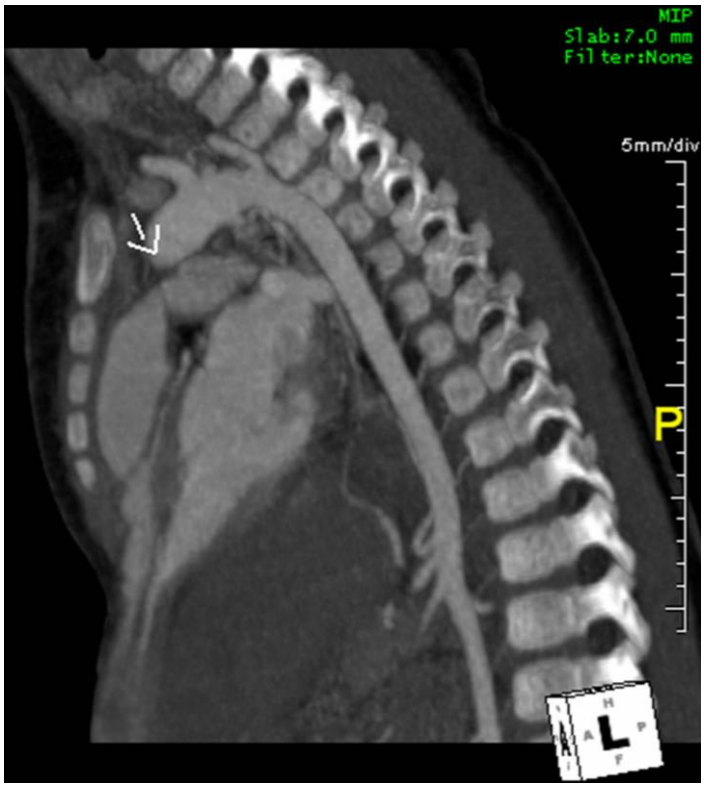


Figure 2: 14-month-old girl with Pentalogy of Cantrell. Axial image at the level of the lower abdomen shows a covered midline abdominal wall defect (arrows) consistent with an omphalocele, which contained part of the left lobe of the liver (Li) along with a portion of the small bowel (SB) and colon (Co). The diverticula (*) were noted anteriorly within the herniated bowel. Along with figure 1 demonstrates the diverticula extending from the inferior margin of the xiphoid process and down the midline abdomen just below the skin surface into the omphalocele. (Technique: Siemens Sensation 64; slice thickness 1.5mm, KvP 120; mAs 80-100; contrast Isovue 370, iopamidol.).



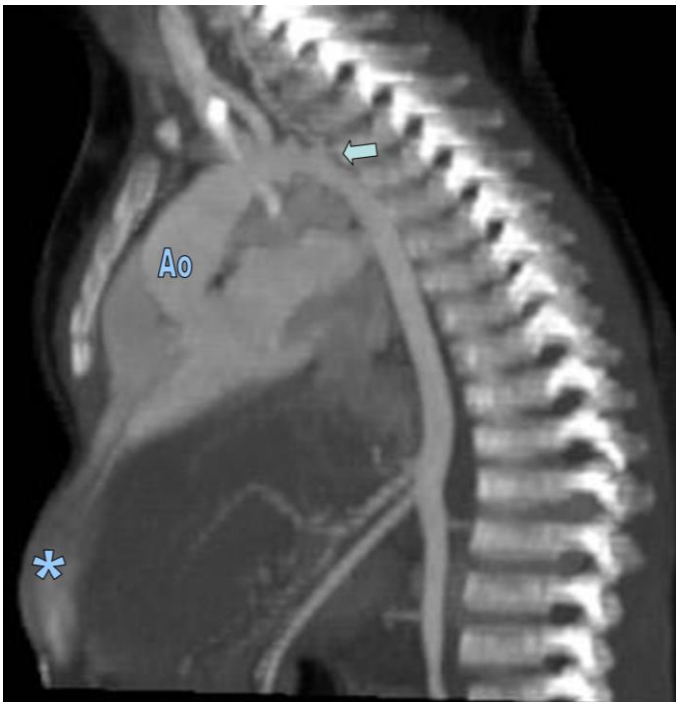


Figure 4: 14-month-old girl with Pentalogy of Cantrell. Oblique Maximum Intensity Projection (MIP) image of the heart shows D-malposition of the great vessels, with the aorta (Ao)located anterior and to the right of the pulmonary artery and left aortic arch with an aberrant right subclavian artery (arrow). There is an elongated left-sided apex extending into the abdominal wall defect (*) (Technique: Siemens Sensation 64; slice thickness 1.5mm, KvP 120; mAs 80-100; contrast Isovue 370, iopamidol.).

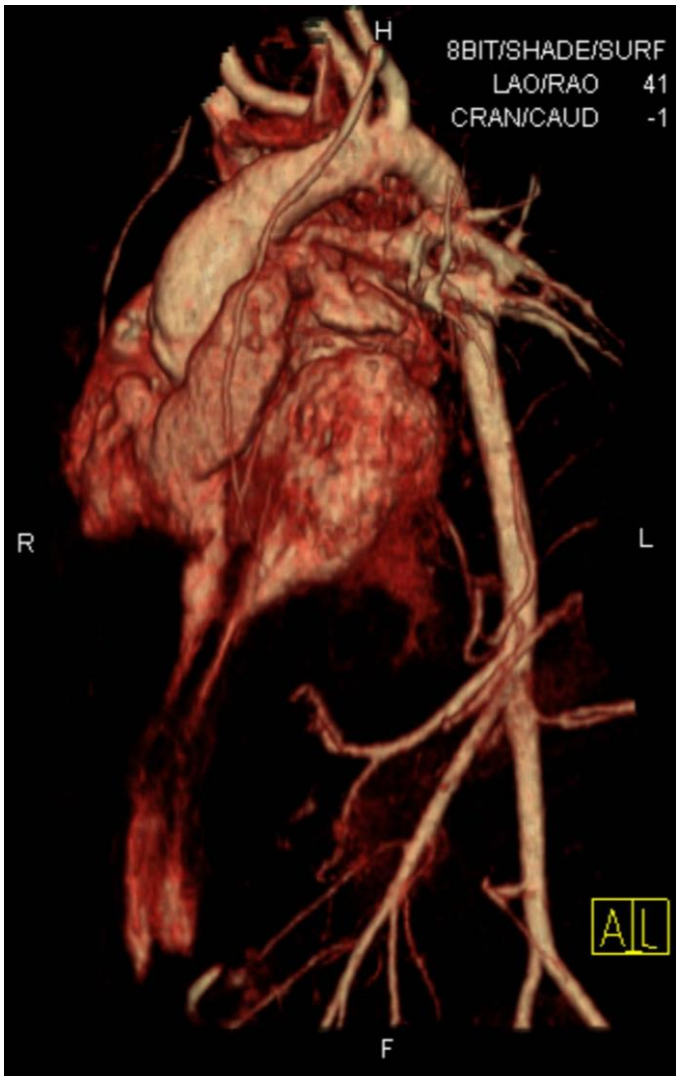


Figure 5: 14-month-old girl with Pentalogy of Cantrell. 3D Volume Rendered image of the heart and thoracoabdominal aorta shows diverticula extending inferiorly from the left and right ventricle into the omphalocele. (Technique: Siemens Sensation 64; slice thickness 1.5mm, KvP 120; mAs 80-100; contrast Isovue 370, iopamidol).

Etiology	A developmental failure of the segmental mesoderm at approximately 14 to 18 days after conception.
Incidence	1 per 65,000 live births
Gender ratio	Male to female 1.35:1
Age predilection	N/A
Risk factors	Suggested X-liked recessive inheritance
Treatment	Surgical correction of the midline developmental defects
Prognosis	Survival of up to 20%, depending on the severity of the intracardiac abnormalities
Findings on imaging	Intracardiac defect, sternal cleft, pericardial defect, diaphragmatic defect, and epigastric omphalocele

Table 1: Summary table for Pentalogy of Cantrell

Complete syndrome	Five defects present: Midline supra-umbilical abdominal defect, defect of the lower sternum, defect of the diaphragmatic pericardium, defect of the anterior diaphragm, and congenital intracardiac abnormality
Probable syndrome	Four defects present, including intracardiac and ventral abdominal wall abnormalities
Incomplete syndrome	With various combinations of defects present, including a sternal abnormality

Table 2: Toyama's proposed classification of Pentalogy of Cantrell

	Pentalogy of Cantrell	**OEIS complex**	**Limb body wall complex**
Malformation	• Intracardiac defect • Sternal cleft Pericardial defect Diaphragmatic defect • Epigastric omphalocele	• Omphalocele–exstrophy of the bladder (cloaca)–imperforate anus–spinal defects	• Severe limb defects and anterior body wall defects (thoracoschisis, bdominoschisis)
Pathogenesis	• Failure of the transverse septum to complete the process of flexion or ventral folding	• Abnormality in the mesenchyme that contributes to the infra-umbilical mesoderm, the urorectal septum, and the lumbosacral somites	• An early amnion disruption sequence in embryogenesis, forming placentocraneal and placentoabdominal adhesions
CT Findings	• Volume and content of the omphalocele • Size of the parietal defect • Identification of cardiac or vascular abnormalities	• Volume and content of the omphalocele • Infraumbilical mass connected to the gut tract • Exstrophied hemi bladder • Spinal defect	• Evaluate thoracoschisis • Differentiate gastroschisis form omphalocele • Facial clefts and/or cranial clefts (with exencephaly)

Table 3: Differential diagnosis table for prenatal detection of abdominal wall defects and multiple midline defects.

ABBREVIATIONS

2D: Two dimensional
3D: Three dimensional
DORV: double outlet right ventricle
MDCT: Multidetector Computed Tomography
MIP: Maximum Intensity Projection
MR: Magnetic Resonance
OEIS: Omphalocele, Exstrophy, Imperforate anus, Spinal defects

KEYWORDS

Pentalogy of Cantrell; MDCT; Ectopia Cordis; Cardiac Malformation; Omphalocele

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