Ectopia Cordis

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Abstract

This case illustrates the critical importance of early prenatal diagnosis, multidisciplinary coordination, and multimodality imaging in the management of ectopia cordis. Advanced imaging not only facilitates accurate diagnosis but also plays a central role in guiding resuscitative strategies and surgical planning. Cardiac MRI provides complementary information to the CT scan with advantages of excellent intracardiac detail (especially for complex anatomy such as double outlet right ventricle, which is commonly seen in ectopia cordis) and functional/flow information.

Keywords: cardiothoracic, cardiac, congenital

Case Summary

A Gravida 1 woman was referred to the maternal-fetal medicine clinic following abnormal findings on routine prenatal US. Sonographic evaluation revealed a structurally abnormal anterior chest wall defect with a beating heart visualized partially outside the thoracic cavity, consistent with ectopia cordis. Further evaluation revealed an omphalocele and a suspected diaphragmatic defect. Subsequent detailed fetal echocardiography demonstrated associated complex intracardiac anomalies, including atrioventricular canal defect with double-outlet right ventricle and pulmonary atresia. A core multidisciplinary team convened to plan the delivery of this high-risk infant, led by maternal-fetal medicine and involving fetal cardiology, neonatology, pediatric cardiothoracic surgery, and pediatric surgery.

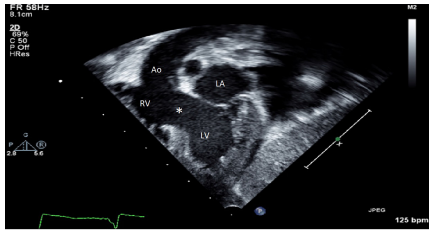
She was born at term with expected SaO_2 of 75-80%, and prostaglandins was initiated for ductal dependent pulmonary blood flow. The prenatal diagnosis was confirmed by transthoracic echocardiography (TTE), and she was palliated with a Blalock-Taussig-Thomas shunt for pulmonary blood flow. During her initial hospitalization, she also underwent omphalocele repair. Other comorbidities included bilateral hydronephrosis of the kidneys.

Planning for further surgical palliation involved multimodality imaging, including cardiac MRI, cardiac catheterization, and CTA, which confirmed a large anterior chest wall defect with cardiac herniation and associated intracardiac defects.

Imaging Findings

The initial postnatal TTE at bedside confirmed the prenatal diagnosis of double outlet right ventricle, with the aorta arising from the right ventricle and a large ventricular septal defect with pulmonary atresia (Figure 1). Cardiac catheterization was done for hemodynamic assessment, which also revealed the dagger-like extension of the left ventricle beyond the sternum (Figure 2) that was also demonstrated with CTA (Figure 3). Cardiac MRI confirmed the moderately dilated left ventricle with its apex below the skin extending beyond the sternum and the right ventricle giving rise to the great arteries (Figures 4, 5).

Figure 1. Postnatal transthoracic echocardiogram 2-D 4-chamber view in systole showing the relative size of ventricles and the large ventricular septal defect (*). The right ventricle gives rise to the aorta. Ao, aorta; LA, left atrium; LV, left ventricle; RV, right ventricle.



Diagnosis

Ectopia cordis.

Ectopia cordis may occur as an isolated anomaly or as part of a broader syndromic spectrum, most notably the pentalogy of Cantrell or limb-body-wall complex. In

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Figure 2. Cardiac catheterization angiogram: left ventricle (10 mL at 15 mL/second via power injection in straight anteroposterior projection). A catheter courses retrograde through the aortic arch into the right ventricle across a ventricular septal defect into the left ventricle. The contrast flows from the left ventricle through a large conoventricular (outlet) ventricular septal defect into the right ventricle and out the aorta. The apex of the left ventricle extends inferiorly coming to a sharp point below the sternum (arrow). The apex is also positioned very anteriorly in the chest wall. Ao, aorta; LV, left ventricle; RV, right ventricle.

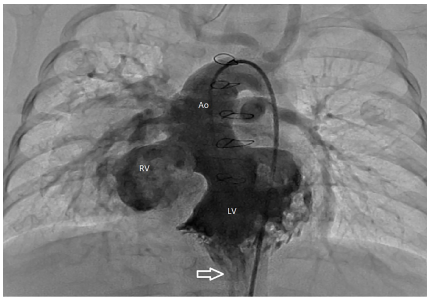
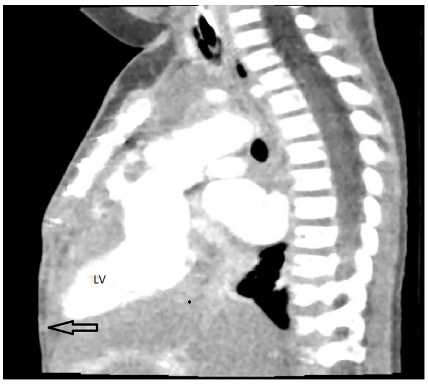


Figure 3. Sagittal contrast-enhanced CT angiogram of the chest demonstrating the left ventricular herniation through the anterior chest wall defect (arrow). LV, left ventricle.



this case, the involvement of the heart, pericardium, diaphragm, sternum, and abdominal wall makes the diagnosis of pentalogy of Cantrell.

Van Allen et al, Russo et al, and many others have outlined various criteria for the diagnosis of limb-body-wall complex, also known as cyllosomas. Limb-body-wall complex is an anomaly consisting of 2 of the following 3 fetal anomalies: (1) thoraco-abdominoschisis or abdominoschisis, (2) limb defects, and (3)

Figure 4. 4-chamber SSFP at end-diastole. The left ventricle apex is just below the skin through a defect in the sternum and ribs (arrow). The right ventricle is normal in size with the left ventricle moderately dilated. The left and right atrium are normal in size. DA, descending aorta; LA, left atrium; LV, left ventricle; RA, right atrium; RPA, right pulmonary artery; RV, right ventricle; SVC, superior vena cava.

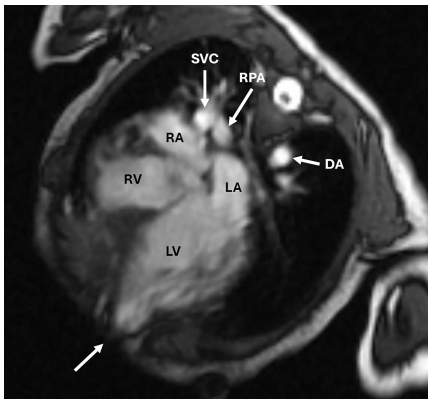
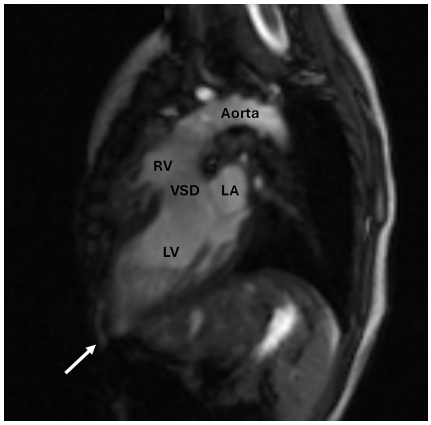


Figure 5. Left ventricular outflow tract SSFP at end-diastole. The left ventricle apex is just below the skin through a defect in the sternum and ribs (arrow). The left ventricle is moderately dilated, and the right ventricle is normal in size. The aorta arises from the right ventricle. There is a large ventricular septal defect. LA, left atrium; LV, left ventricle; RV, right ventricle; VSD, ventricular septal defect.



cranio-facial abnormalities, such as cleft lip/palate, encephalocele, or exencephaly.

Discussion

Ectopia cordis is a rare and severe congenital malformation characterized by partial or complete extrusion of the heart outside the thoracic cavity, resulting from failure of midline mesodermal fusion during early embryogenesis.[1-4] It is frequently associated with intracardiac defects and extracardiac anomalies such as omphalocele and diaphragmatic hernia, contributing to its historically poor prognosis.

In this case, the pulmonary atresia necessitated early palliative cardiac surgery to provide pulmonary blood flow. The prenatal diagnosis was crucial for preparing for immediate postnatal resuscitation and management, including surgical planning. However, until 1981, ectopia cordis could only be diagnosed upon delivery, and prior to 1988, there were no reported cases of prenatal diagnosis of this malformation before the 3rd trimester of gestation.[5]

In the modern day, ectopia cordis can be diagnosed as early as 10 weeks of gestation, but it is typically diagnosed at the beginning of the 2nd trimester via a fetal echocardiogram, which remains the primary diagnostic tool.[5,6] The combination of fetal echocardiography and MRI enables precise prenatal characterization, facilitating delivery planning, resuscitation preparedness, and multidisciplinary coordination. While fetal echocardiography remains the cornerstone of diagnosis, fetal MRI provides superior delineation of extracardiac anatomy and spatial relationships but may not be routinely available at all centers.

Postnatally, TTE plays a central role in delineating intracardiac anatomy and assessing hemodynamics, including ventricular function and associated structural defects such as ventricular septal defects. Cardiac MRI provides enhanced spatial resolution and tissue characterization, allowing for improved visualization of extracardiac relationships and more comprehensive functional assessment. CTA is particularly valuable in preoperative planning, as it offers high-resolution delineation of both

cardiovascular and extracardiac anatomy. In addition, CTA enables accurate assessment of thoracic cavity dimensions and lung volumes, which are critical in determining the feasibility, timing, and surgical approach for repair in patients with ectopia cordis.[7] Despite advances in prenatal diagnosis and perioperative care, outcomes remain guarded and are highly dependent on the extent of cardiac exposure, associated anomalies, and feasibility of surgical reconstruction. Overall, the survival rate is approximately 10% depending on gestational age and associated cardiac and extracardiac anomalies. This case highlights the evolving role of multimodality imaging in optimizing early management strategies for this rare but life-threatening condition.

Conclusion

This case illustrates the critical importance of early prenatal diagnosis, multidisciplinary coordination, and multimodality imaging in the management of ectopia cordis. Advanced imaging not only facilitates accurate diagnosis but also plays a central role in guiding resuscitative strategies and surgical planning. Cardiac MRI provides complementary information to the CT scan with advantages of excellent intracardiac detail (especially for complex anatomy such as double outlet right ventricle, which is commonly seen in ectopia cordis) and functional/flow information.

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