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Transcatheter Atrial Septal Defect Closure in Unrepaired Pentalogy of Cantrell with Ectopia Cordis: First Reported Case

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ABSTRACT

Background: Pentalogy of Cantrell (PC) with ectopia cordis (EC) is rare and often fatal. Intracardiac anomalies, particularly secundum atrial septal defect (ASD), are common, occurring in ~80% of cases. However, percutaneous ASD closure prior to chest wall reconstruction has not been reported.

Case: A 14-month-old, 6.9-kg girl with unrepaired PC-EC, with history of recurrent respiratory infections, and failure to thrive had a large secundum ASD (~11–12 mm) with flail IVC rim. Given high surgical risk, transcatheter closure was attempted. The unique challenges included (i) poor echocardiographic visualization of defect and (ii) extreme cardiac mobility due to deficient sternum causing repeated device prolapse. Using a left-atrial (LA) roof technique, a 14-mm Lifetech ASD occlude device was successfully deployed a gentle wiggle maneuver assessed on bed-side echocardiography confirmed optimal device position before release.

Outcome: Three-month follow-up showed complete occlusion, no cardiac complications, weight gain (~4 kg), and resolution of infections.

Conclusion: Percutaneous ASD closure may serve as a bridge to definitive repair in selected PC-EC patients. The case underscores the unique challenges of transcatheter ASD closure in setting of a deficient chest wall and highlights the importance of operator familiarity with alternative device deployment techniques.

Keywords: Ectopia cordis; Pentalogy of Cantrell; Atrial septal defect; Transcatheter closure; Left atrial roof technique; Chest wall defect

INTRODUCTION

Ectopia cordis (EC) is an exceptionally rare congenital anomaly, with an estimated incidence of only 5.5-7.9 per million live births and accounting for merely 0.5–0.8% of all congenital heart defects [1,2]. Intracardiac malformations are extremely common in PC-most series report ~80% of patients having some congenital heart disease [1-3].

Ectopia cordis (EC), in which the heart lies partially or completely outside the thoracic cavity, represents the most dramatic manifestation of PC and carries a reported survival of <5% [3]. Management generally involves staged surgical internalization of the heart and reconstruction of the chest wall. When significant intracardiac shunts such as atrial septal defect (ASD) coexist, cardiopulmonary bypass during early reconstructive surgery becomes necessary, increasing the perioperative risk in already frail infants [4]. A hybrid approach, wherein an intracardiac lesion can first be addressed by transcatheter intervention followed by staged surgical reconstruction can hypothetically improve the outcomes in these otherwise high risk patients.

Transcatheter ASD closure is standard of care for significant secundum defects with suitable anatomy, but is usually performed after 2 years of age [5] and has never been reported in an untreated PC-EC patient. Only one case has been published where transcatheter ASD closure was performed after prior chest wall reconstruction [6]. Therefore, the unique challenges of transcatheter device closure in unrepaired EC remain unreported. We present the first successful percutaneous closure of a large secundum ASD in a child with unrepaired EC, highlighting the unique anatomic and technical challenges encountered.

CASE PRESENTATION

Clinical history

A 14-month-old female infant (weight 6.9 kg, less than 3rd percentile) was referred with a postnatal diagnosis of PC with EC. The mother reported recurrent lower respiratory tract infections and diaphoresis while crying. Growth had been poor since birth. Physical examination revealed that lower end of the sternum was absent. The point of maximal apical impulse was located at the inferior border of the xiphoid process. The upper abdominal musculature beneath this pulsating area was deficient, although the overlying skin was intact, and connected to the umbilicus by a cord-like structure (Video 1-6). Oxygen saturation was 99% on room air.

Investigations

Chest radiography showed gross right heart enlargement with pulmonary plethora (Figure 1). Standard transthoracic echocardiographic (TTE) windows were suboptimal because of the heart's extrathoracic location; modified off-axis subcostal views revealed a large ostium secundum ASD measuring approximately 11 mm with a flail inferior vena cava (IVC) margin, dilated right atrium (RA) and right ventricle, mild pulmonary artery hypertension with no additional structural defects (Figure 2 a and b).

Given the patient's severe malnutrition and recurrent respiratory infections, the multidisciplinary team considered primary chest wall reconstruction along with ASD patch closure on cardiopulmonary bypass to be extremely high risk. A strategy of initial percutaneous ASD closure to reduce pulmonary overcirculation and improve nutritional status was therefore.

Procedure

After obtaining informed consent from parents, the procedure was performed under conscious sedation (using intravenous midazolam and fentanyl) and local anesthesia. Femoral venous access was obtained using a 6F sheath that was later upgraded to 7F. Intraprocedural TTE was performed using modified views.

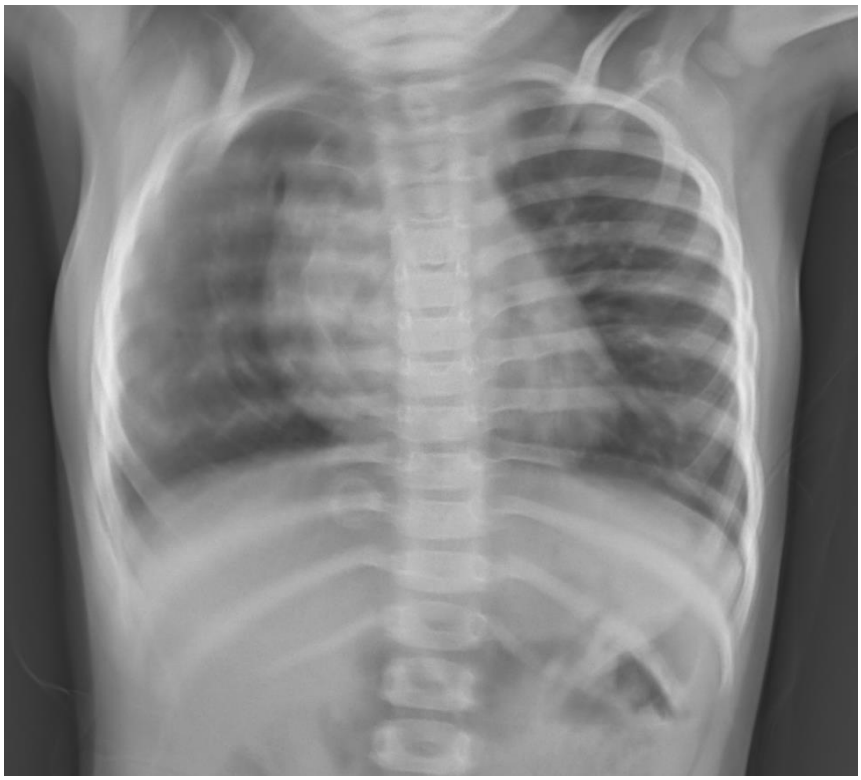

Figure 1: Chest Xray in postero-anterior projection: Right heart enlargement and prominent pulmonary vascular markings can be noted.

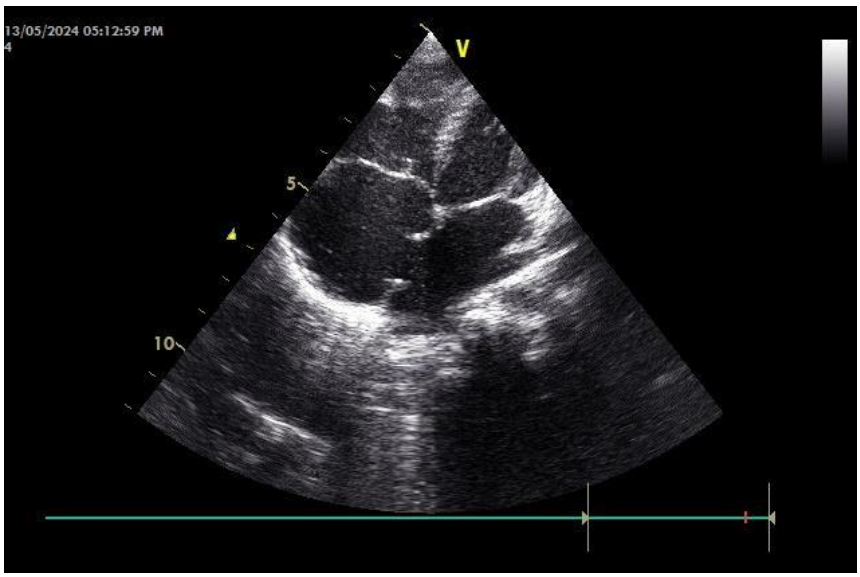

Figure 2A: Modified sub-costal coronal view: Showing os-ASD with thin posterior margin.

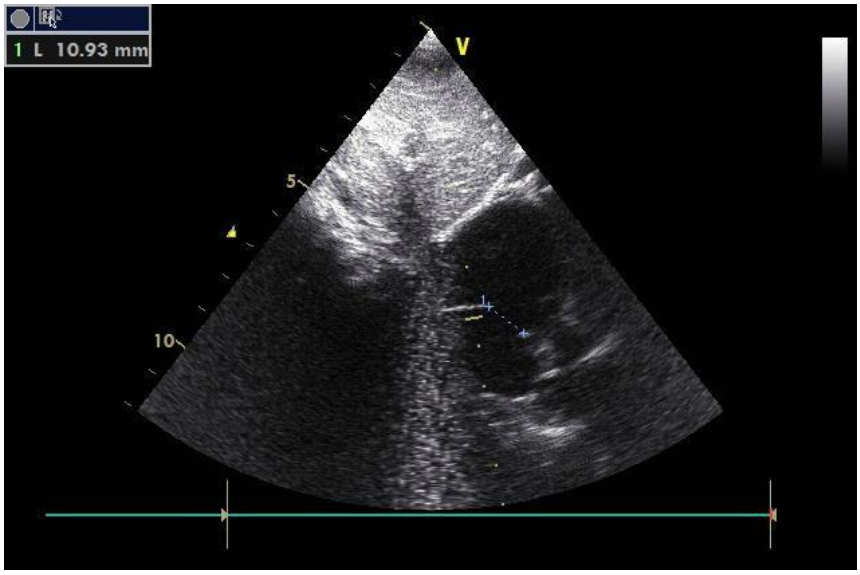

Figure 2B: Modified sub-costal bicaval view: Showing 10.93mm os-ASD with flail IVC margin.
Abbreviations: os-ASD: ostium secundum atrial septal defect, IVC: inferior venal cava

Since echocardiographic views were sub-optimal for defect sizing hence balloon sizing of the atrial septal defect was done that revealed a stop-flow diameter of approximately 13.5 mm. (Figure 3). A 14mm Lifetech Cera ASD occluder device was chosen that can be delivered through a 7F SteerEase Lifetech delivery system. We deliberately avoided any over-sizing of the device in consideration of an underweight small child.

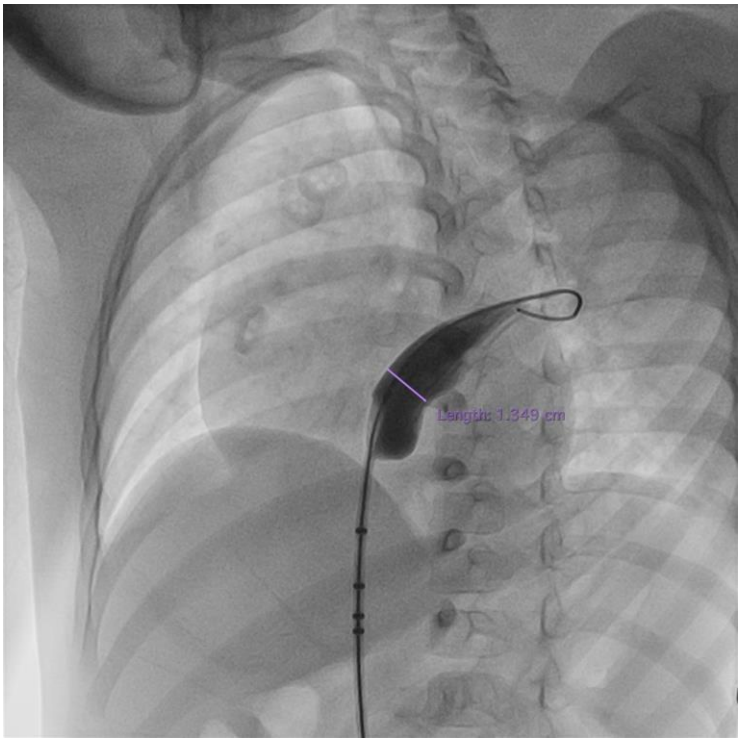

Figure 3: Balloon interrogation suggesting an ASD diameter of 13.49mm.

An initial attempt at device deployment with right and left upper pulmonary vein approach resulted in repeated prolapse of device into the right atrium because of unstable delivery system (Video 2). The absence of a stabilizing median musculoskeletal chest wall allowed the heart to swing freely, transmitting large excursions to the delivery sheath. The unstable platform made it difficult to align the left atrial (LA) disc with the atrial septum.

Multiple maneuvers were attempted, including varying the sheath curve and clockwise torquing of sheath without success. Finally, an LA roof technique was employed (Video 3): the sheath was advanced to the LA roof to create a more stable rail, the LA disc was anchored by deploying it against the atrial roof, followed by deployment of RA disc on right side of septum. This maneuver allowed successful deployment of the ASD occluder.

However, before releasing an ASD device, it is our general practice to look for optimal disc separation in left anterior oblique -cranial (LAO-CRAN) projection. However in this case, the LA and RA discs seemed to approximate each other at the lower pole of device in LAO-CRAN view. (Video 4) To rule out missing the inferior ASD margin, a wiggle was then performed with simultaneous echocardiography (ECHO) that showed postero-inferior margin to be well captured by the device (Video 5). The misleading fluoroscopic appearance was likely due to thin and flail postero-inferior margin giving false impression of both discs "touching" one-another. The device was then released.

Immediate echocardiography confirmed stable device position with complete occlusion of the defect and no pericardial effusion or arrhythmia. The infant was then shifted to cardiac care unit for recovery.

Outcome and Follow-Up

Post-procedure echocardiography at 24 hours demonstrated satisfactory device position and reduced right atrial dimensions (Video 6). The patient was discharged on dual antiplatelet therapy for three months.

At three-month follow-up, the child had gained 4 kg and remained free of respiratory infections. She is currently awaiting elective chest wall and abdominal wall reconstruction.

DISCUSSION

Cantrell defined PC as a constellation of five defects: an intracardiac structural anomaly, displacement of the heart, absence of the diaphragmatic pericardium, a defect of the lower sternum, and a midline supra-umbilical abdominal wall deficiency. Owing to the frequent presence of cardiac ectopia, it is often described as a form of thoracoabdominal EC [1,2]. Intracardiac malformations are extremely common in PC-most series report ~80% of patients having some congenital heart disease [7,8].

The prevailing consensus is that management of PC requires an individualized approach, determined by the anatomical complexity of the thoracoabdominal defects and the associated intracardiac lesions. The presence of a major intracardiac shunt lesions like ASD can further complicate management decisions and contributes to the overall poor prognosis [9]. In our case, poor nourishment status of the child and repeated respiratory infections due to a large ASD prompted us to consider transcatheter closure prior to surgical reconstruction.

The unique anatomic situation in EC posed several challenges during the procedure:

1. Imaging limitations: Transthoracic ECHO required non-standard imaging planes. Accurate assessment of defect size was therefore technically demanding. We attempted to avoid complications associated with general anesthesia in this otherwise frail child and hence trans-esophageal ECHO could not be used. Instead, balloon sizing of the defect was done for choosing the appropriate device size.
2. Unstable delivery system: In normal anatomy, the thoracic cage provides counter-pressure that stabilizes the heart during cardiac cycle. In ectopia cordis, the heart lies in a relatively unrestricted space and moves markedly with respiration and cardiac contraction. This exaggerated motion translated into significant instability of the long delivery sheath, causing repeated prolapse of the device during standard deployment.
3. Fluoroscopic projection in the conventional LAO-CRAN view was misleading as it suggested device malposition prior to release. This was due to thin and flail postero-inferior margin and optimal device position was reconfirmed with wiggle maneuver visualized on bed-side ECHO.

Operator familiarity with alternative deployment maneuvers was critical for success. The LA roof technique, previously described in difficult atrial septal anatomies, provided the extra stability needed to deploy the device securely [10].

Galeczka et al. reported the only previously published case of ASD device closure in a PC–EC patient, but in their report chest wall reconstruction preceded transcatheter closure [6]. McMohan have described the only case of transcatheter right ventricular outflow tract stenting in unrepaired EC [11]. Our patient underwent percutaneous closure before any surgical repair, thereby revealing the unique challenges for an interventionist in this rare embryological condition.

Early percutaneous ASD closure may offer a bridge to definitive chest wall reconstruction in selected high-risk PC–EC patients. By reducing pulmonary overcirculation and recurrent respiratory infections, it can improve nutritional status and overall surgical candidacy.

This case, to our knowledge, is the first report of transcatheter ASD closure in a child with unrepaired PC-EC.

CONCLUSION

Percutaneous ASD device closure is feasible even in the challenging anatomy of unrepaired PC with EC. The absence of a chest wall creates marked cardiac mobility, demanding improvisation in imaging and device deployment technique during transcatheter closure of ASD. In carefully selected patients who are poor candidates for immediate reconstructive surgery, this staged approach may significantly improve outcomes.

REFERENCES

1. Toyama WM. Combined congenital defects of the anterior abdominal wall, sternum, diaphragm, pericardium, and heart: a case report and review of the syndrome. Pediatrics. 1972;50(5):778-792.
2. Cantrell JR, Haller JA, Ravitch MM. A syndrome of congenital defects involving the abdominal wall, sternum, diaphragm, pericardium, and heart. Surg Gynecol Obstet. 1958;107(5):602-614.
3. Pius S, Abubakar Ibrahim H, Bello M, Bashir Tahir M. Complete ectopia cordis: a case report and literature review. Case Rep Pediatr. 2017;2017:1858621.
4. Liao J, Huang H, Li X. Surgical treatment of neonatal Cantrell pentalogy: a case report and literature review. AME Case Rep. 2023;7:22.
5. Stout KK, Daniels CJ, Aboulhosn JA, Bozkurt B, Broberg CS, Colman JM, et al. 2018 AHA/ACC guideline for the management of adults with congenital heart disease: a report of the American College of Cardiology/American Heart Association Task Force on Clinical Practice Guidelines. Circulation. 2019;139(14):e698-e800.
6. Galeczka M, Fiszer R, Knop MT, Smerdzinski S, Szkutnik M, Bialkowski J. Successful atrial septal defect transcatheter closure in a patient with pentalogy of Cantrell and ectopia cordis. Postepy Kardiol Interwencyjnej. 2019;15(2):247-250.
7. Medina-Escobedo G, Reyes-Mugica M, Arteaga-Martinez M. Ectopia cordis: autopsy findings in four cases. Pediatr Pathol. 1991;11(1):85-95.
8. Fernández MS, López A, Vila JJ, Lluna J, Miranda J. Cantrell's pentalogy. Report of four cases and their management. Pediatr Surg Int. 1997;12(5-6):428-31.
9. Morales JM, Patel SG, Duff JA, Villareal RL, Simpson JW. Ectopia cordis and other midline defects. Ann Thorac Surg.2000;70(1):111-4.
10. Jung SY, Choi JY. Transcatheter closure of atrial septal defect: principles and available devices. J Thorac Dis.2018;10(Suppl 24):S2909-S2922.
11. McMahon CJ, Walsh KP. Transcatheter right ventricular outflow tract stent implantation in a child with pentalogy of Cantrell, double outlet right ventricle, and severe pulmonary stenosis. Catheter Cardiovasc Interv. 2013;82(7):1164-7.