Case Report

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Ectopia Cordis: A Rare Congenital Heart Disease

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Keywords:
Congenital heart disease, Ectopia cordis, Single ventricle.

Abstract:

Three days old female presented with beating heart outside the thoracic cavity. This is a rare congenital abnormality associated multiple abnormalities. Patient came with features of infection. To diagnose routine blood investigation along with echocardiography and CT angiogram of heart,great vessel and abdomen were done. She was diagnosed as functionally single ventricle with hypoplastic branch pulmonary artery with malposed great artery with anomalous pulmonary venous drainage. Thisis lethal variety of ectopic cordis. Our patient had no adequate space in mediastinum to replace heart and she was in septicemia, so managed medically. Patient expired at her 6thday due to sepsis and intracardiac complex congenital heart disease.

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Case Report:

Three days old female born at 37 wks of gestation, weighing 2.34kg (low birth weight) delivered by Cesarean section. The baby had thoracic wall defect with beating heart outside the mediastinum. There was no history of consanguineous marriage, antenatal infection, intake of any teratogens, drugs or exposure to radiation, etc. in antenatal period.

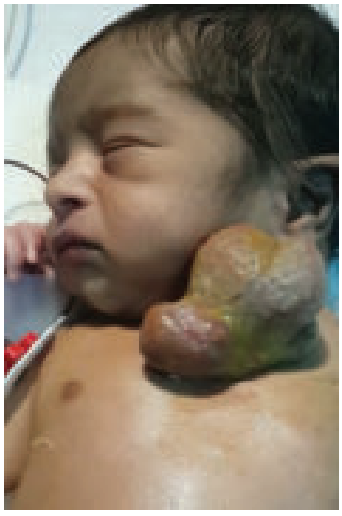

Fig-1: *The baby had thoracic wall defect with heart outside the mediastinum.*

On examination she was found with respiratory distressed (Respiratory rate- 25/min, shallow breathing) with cyanosis (SPO2-60%), bradycardia (pulse- 60/min, regular) with poor reflex, vesicular breath sound with no added sound & S1, S2 normal with ejection systolic murmur.Echocardiography was challenging, done with aseptic way. Finding was Functional single ventricle with atrial (ASD) & ventricle septal defect (VSD) with nonvisualization of pulmonary tree.

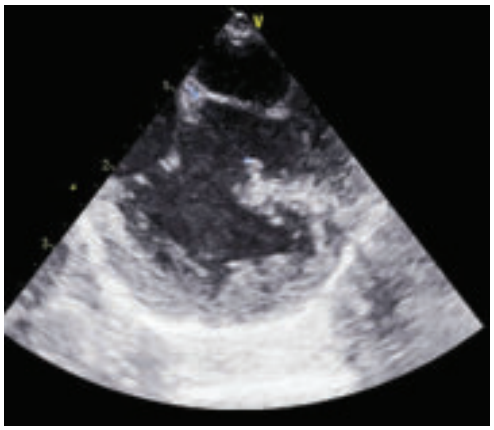

Fig.-2: *Echocardiographic apical four chamber showing large inlet VSD almost functionally singleventricle.*

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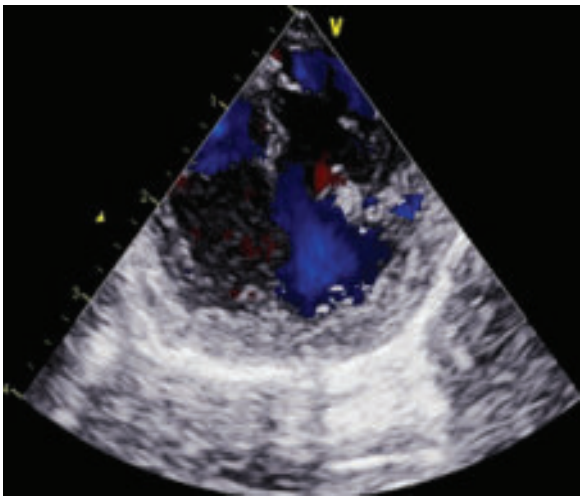

Fig.-3: *EchocardiographicApical four chamber showing large Inlet VSD with bidirectional shunt.*

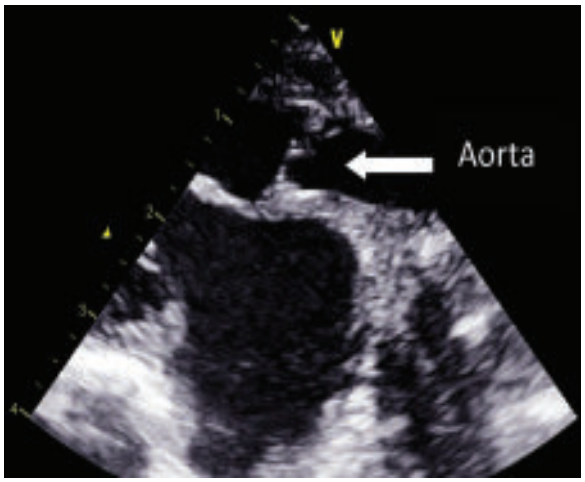

Fig.-4: *Echocardiographic modified view showing single trunk (aorta)arising from left ventricle.*

Echo findings confirmed by CT angiogram and found additional finding that was essential for surgical decision. Pulmonary artery & Veins were not well delineated in echocardiography. CT angiography showed functional single ventricle due to large ventricular septal defect with small left ventricle with severely hypoplastic branch pulmonary artery with malposed great artery with anomalous pulmonary venous drainage to vertical vein which drained to left atrium with large atrial septal defect.

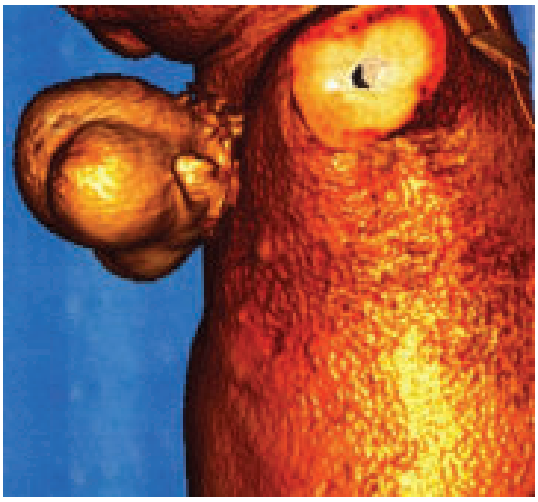

Fig.-5: *Defect in thoracic cavity & heart lies perpendicular to body.*

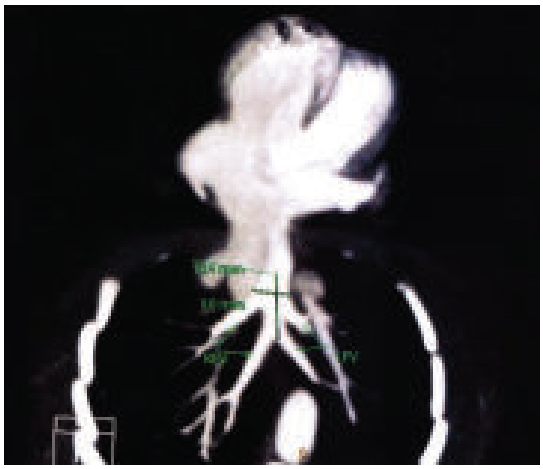

Fig-6: *CT angiogram showing pulmonary veins draining to left atrium through a vertical vein.*

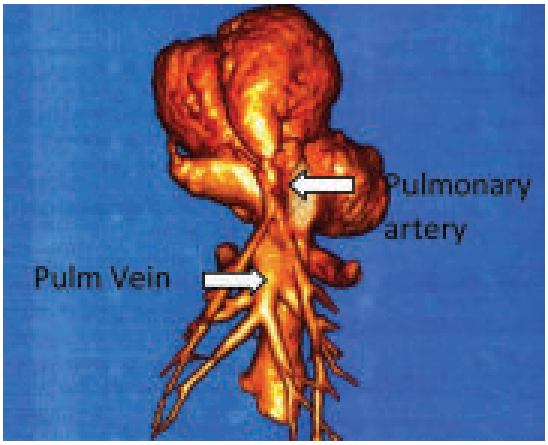

Fig.-7: *CT angiogram showing severely hypoplastic pulmonary artery with its confluent branch which arising from right ventricle.*

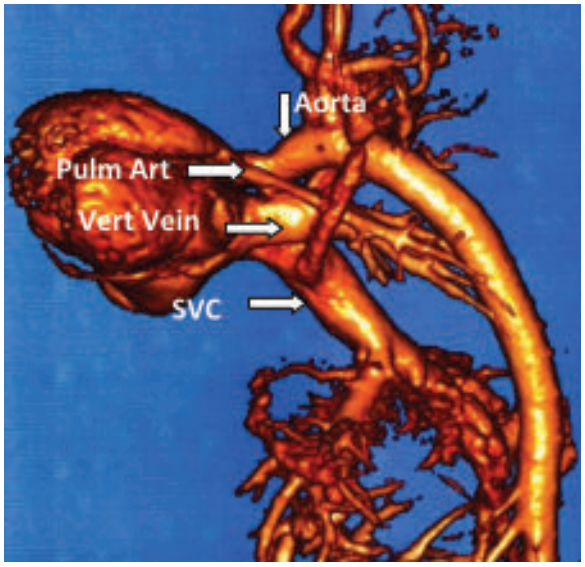

Fig.-8: *CT angiogram, lateral view showing great artery relation. Pulmonary artery and aorta side by side relation. Vertical vein (pulmonary venous confluence) and SVC is dilated and lies posteriorly.*

Surgical repositioning of heart is difficult for this patient because there was barely any space in mediastinum. No extra cardiac anomaly found. Patient was kept in temperature regulated (35°C) incubator. Initial management included covering of the heart with sterile-saline soaked dressing, systemic antibiotics and supportive treatment. The neonate had progressive respiratory distress, hypothermia & lethargy. With all support failed to resuscitate the child &had cardiorespiratory arrest before any surgical intervention could be undertaken.

Discussion:

Ectopia cordis is an extremely rare congenital malformation which is a Latin word where the heart is located partially or totally outside the thoracic cavity.[1]The ûrst case was reported by Neil Stenonis, who described a child with an open sternal line and protruding heart, liver and spleen.[2] In 1706, Haller and Martý´nez separately reported cases of ectopia cordis.[3]It is generally a sporadic malformation, with reports linking it to chromosomal abnormalities like trisomy 18, Turner syndrome, 46,XX, and 17q+.[4-8]Ectopia cordis has also been attributed to intrauterine drug exposure in animal models.[9,10]The occurrence prevalence is estimated to be between 5.5 and 7.9/million live births.[4,11]

There are four types of ectopia cordis identified (1) thoracic (65%), (2) thoraco-abdominal (20%), (3) abdominal (10%), and (4) cervical(5%).[3]The thoracic variety was our case has been reported to have worst prognosis with <5% surviving beyond the first month of life.[12]

Ectopia cordis results from the failure of migration of lateral mesoderm into the midline. The most common associations include sternal defect, pericardial defect, cardiac anomalies and abdominal wall defect, commonly referred to as Pentology of Cantrell.[13] In this newborn, there were no other features of Pentology of Cantrell.

The most common forms of intra-cardiac defects in ectopia cordis include common atrium, AV septal defect, ventricular septal defect, outflow tract anomalies and single ventricle. Our patient was diagnosed with single ventricle with large VSD and ASD with malposed great artery with severely hypoplastic pulmonary tree.

Ectopia cordis can be diagnosed by routine prenatal ultrasonography as early as in 10-12 weeks of pregnancy.[14,15] Of those not diagnosed antenataly, most result in stillbirth or die shortly after birth due to their frequent association with intrinsic cardiac and other congenital defects.[15,16]Newborns with this complex and life threatening deformity require intensive care right from birth. They require immediate resuscitation and coverage to the exposed heart and viscera with saline-soaked gauze pads wrapping to prevent desiccation and heat loss. For this, Harrison et al. have reported use of adherent plastic drapes also.[17]Thoracic Ectopia cordispresents a formidable surgical challenge. In recent reviews of the literature the reported survival of this variety after birth averages 36 h; intracardiac defects were associated in 80.2% of the cases, and all unoperated patients died.[18] Besides intrinsic cardiac defects, the increased morbidity in these patients may be attributed to the abnormal course, length and positioning of great vessels of the heart making them prone to kinking and further compromising circulation, as also must have been in our case. During surgical closure, in most of the cases the

thoracic cavity is small with little mediastinal space for the heart. Attempts to close the chest wall often result in intolerable haemodynamic embarrassment secondary to kinking of the great vessels possibly due to their long length and abnormal course, or compression of the heart. Therefore, a staged repair is often necessary. The strategy for repair is divided in two stages: (1) urgent soft tissue coverage and haemodynamic palliation if necessary; and (2) intracardiac repair with concomitant chest wall reconstruction and reduction of the heart into the thoracic cavity.

The overall surgical objective of Ectopia cordis (all variety) management includes: 1) closure of the chest wall defect (either by doing primary chest wall closures or by using bone/cartilage as tissue graft or artificial prosthesis like acrylic plaques, marlex mesh), 2) closure of the sternal defect, 3) repair of the associated omphalocele, 4) placement of the heart into the thorax, 5) repair of the intracardiac defect.

The first attempted repair of Ectopia cordis was performed in 1925 by Cutler and Wilens.[19]Koop in 1975 achieved the first successful repair of thoracic Ectopia cordis in two stages.[20]Amato et al reported successful single stage repair of thoracic EC in 1995.[21] Conclave et al. at Brazil reported a successful repair of uncomplicated Ectopia cordis in June 2007.[22]

Conclusion:

Prognosis depends upon the variety of ectopia cordis, space in thoracic cavity, alignment & length of great artery,associated intracardiac and extracardiac anomaly. Our case was having bad prognostic factor along with sepsis. But we have to keep in mind it can be treatable & there are several reports where patient were survived by stage repair. Antenatal ultrasonography plays an important role in detecting such anomalies which may be important for planning its further management.

Conflict of Interest - None.

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