Rare disease

Thoracic ectopia cordis

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Ectopia cordis is defined as complete or partial displacement of the heart outside the thoracic cavity. It is a rare congenital defect in fusion of the anterior chest wall resulting in extra thoracic location of the heart. Its estimated prevalence is 5.5–7.9 per million live births. The authors had one such case of a 15-h-old full-term male neonate weighing 2.25 kg with an externally visible, beating heart over the chest wall. The neonate had difficulty in respiration with peripheral cyanosis. Patient died of cardiorespiratory arrest before any surgical intervention could be undertaken in spite of best possible resuscitative measures.

BACKGROUND

The interesting and curious presentation of the case associated with infrequent occurrence led us to report this case.

CASE PRESENTATION

A 15-h-old, normal vaginally delivered full-term male neonate, weighing 2.25 kg was brought with an externally visible, beating heart over the chest wall and difficulty in respiration. There was no history of consanguineous marriage, infection, intake of any teratogens, drugs or exposure to radiation, etc. in antenatal period. The antenatal period was uneventful, though no ultrasonography was done. There was no family history of any such or related congenital abnormality. Examination revealed peripheral cyanosis with heart rate-150/min, respiratory rate-76/min and SpO₂-88%. The lower half of sternum was bifid, with 5 cm inter-ridge distance. The heart, lying outside the thoracic cavity was devoid of pericardium (figures 1 and 2; video 1), with its apex pointing anteriorly. Baby also had supraumbilical omphalocele with umbilical cord attached at the lower part of the defect.

INVESTIGATIONS

Investigations: haemoglobin-12 g%, total leucocyte count-8200/cumm, differential leucocyte count: P-48%, L-42%, E-4%, M-2%, B-0%. Platelet count-2.3 lakhs/cumm. Blood group-B+ve. Serum bilirubin: T-1.1 mg%, D-0.4 mg%, I-0.7 mg%. Serum Na-146 mmol/l, serum potassium-3 mmol/l.

X-ray chest showed crowding of ribs with bifid lower sternum. Echocardiography showed ventricular septal and pericardial defects.

DIFFERENTIAL DIAGNOSIS

- ▶ Pentalogy of Cantrell.
- ▶ Amniotic band syndrome.
- ▶ Cyllosomas.

TREATMENT

Patient was kept in temperature regulated (35°C) incubator in propped up posture. Initial management included

covering of the heart and omphalocele with sterile-saline soaked dressing, systemic antibiotics and supportive treatment. The neonate had progressive respiratory distress, hence intubated, and shifted on mechanical ventilation. Inotropic drugs infusion was started to support the heart and maintain systemic blood pressure. Despite treatment, the child remained hypotensive with deteriorating oxygen saturation levels and blood gas analysis showing progressive acidemia and dyselectrolytemia. Efforts to correct acidosis and electrolyte imbalance were made, but the neonate did not improve and had cardiorespiratory arrest before any surgical intervention could be undertaken.

Video 1 Video shows externally visible beating heart lacking any skin or pericardial covering along with chest wall defect, omphalocele, inferior insertion of umbilical cord.

OUTCOME AND FOLLOW-UP

The neonate succumbed before any surgical intervention.

DISCUSSION

Ectopia cordis (EC) was first observed 5000 years ago¹ Haller first described the term EC in 1706. EC is defined as complete or partial displacement of the heart outside the thoracic cavity. It is a rare congenital defect in fusion of the anterior chest wall resulting in extra thoracic location of the heart. Its prevalence has been estimated to be 5.5–7.9 per million live births.^{2–3} It is generally a sporadic malformation, with reports, linking it to chromosomal abnormalities like trisomy 18,^{4–5} Turner syndrome and 46, XX, 17q+.⁶ In our case, the child appeared to have long philtrum and low implant of the ears but on thorough clinical examination no definitive association with any syndrome could be documented and karyotyping to establish any genetic associations could not be done.

Depending upon the location of the heart, it can be classified into five types: cervical (5%), cervicothoracic and thoracic (65%), thoracoabdominal (20%) and abdominal (10%).⁷ The combination of thoracoabdominal EC, lower sternal defect, anterior diaphragmatic hernia, midline supraumbilical defect

Figure 1 Patient with externally visible heart, no skin or pericardial covering is present over the heart, omphalocele present inferiorly to ectopic heart with umbilical cord inserted at the inferior end of abdominal wall defect.

along with pericardial and intracardiac defects constitute the Pentology of Cantrell.⁸⁻⁹ The thoracic variety, as the present case was, has been reported to have worst prognosis with <5% surviving beyond the first month of life.¹⁰

EC can be diagnosed by routine prenatal ultrasonography as early as in 10-12 weeks of pregnancy.¹¹⁻¹² Of those not diagnosed antenatally, most result in stillbirth or die shortly after birth due to their frequent association with intrinsic cardiac and other congenital defects.¹²⁻¹³

The development of the ventral body wall begins by eighth day of embryonic life with differentiation and proliferation of mesoderm followed by its lateral migration. The heart originally develops in a cephalic location and reaches its definitive position by the lateral folding and ventral flexing of the embryo at about 16th–17th day. Midline fusion and formation of the thoracic and abdominal cavities is complete by the 9th embryonic week.¹⁴⁻¹⁵ Complete or incomplete failure of midline fusion at this stage result in disorders varying from isolated EC to complete ventral evisceration. Genesis of EC has not been fully explained, although several theories have been offered.¹⁶ Popular theories are early rupture of the chorion and/or yolk sac, and amniotic band syndrome.¹⁷⁻¹⁹

The amnion rupture theory states that during early embryonic development, the amnion surrounding the embryo ruptures, and stringy, sticky, fibrous bands of amnion becomes ‘entangled’ with the forming embryo, and causes a disruption in the developing parts of the fetus which may lead to various deformities like EC, midline sternal cleft, frontonasal dysgenesis, a midfacial cleft, limb deformities etc. The spectrum of defect corresponds to the timing of its rupture. The findings in the literature suggest that its rupture in the third week of gestation causes an arrest of cardiac descent which may be the cause of such defects. EC with amniotic bands appears to have aetiology distinct from isolated EC. This suggests several different aetiologies for EC.¹⁷⁻¹⁸ EC has also been attributed to intrauterine drug exposure in animal models.²⁰⁻²¹

Cyllosomas, another name for limb-body wall complex is defined as anomaly consisting of two of the following three fetal anomalies: a) thoraco-abdominoschisis (opening extends from the chest through the abdomen) or

Figure 2 Lateral aspect of the patient.

abdominoschisis (opening starts at a lower point and only extends through the abdomen), b) limb defects, c) craniofacial defects- cleft lip/palate, encephalocele, exencephaly, etc. and others.²²

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.²³ Thoracic EC presents 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.²⁴ 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 EC (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 EC was performed in 1925 by Cutler and Wilens.²⁵ Koop in 1975 achieved the first successful repair of thoracic EC in two stages.²⁶ Amato *et al* reported successful single stage repair of thoracic EC in 1995.²⁷ Cristofor Wall operated by Koop in August-1975 is the oldest live pt of EC. Other surviving operated cases which could be traced on literature search are:

- ▶ Aytag and Sayam successfully operated partial EC in 1976.²⁸
- ▶ Dobell *et al*, reported two stage correction¹⁶
- ▶ Kim KA *et al*, – Los Angeles – 1996, did two – stage operation.²⁹
- ▶ Morales *et al* – reported successful 2-stage repair of EC on female baby.³⁰
- ▶ Khaled *et al* from France reported successful single stage repair in 2003.³
- ▶ Gonçalves *et al* at Brazil reported successful repair of uncomplicated EC in June 2007.³¹

Hence it may be concluded that EC is a lethal anomaly requiring prompt medical and surgical interventions. Surgery on these patients with life-threatening complex intracardiac anomalies, owes the only chance of survival, which should still be attempted despite heretofore poor outcomes. Although the available literature is scant, but with the advance in all aspects of medicine, the number of patients who undergo successful surgical repair and survive should steadily increase, with our combined efforts to understand and formulate treating protocol of this extremely rare anomaly.

Learning points

- ▶ Success of repair is dictated by the presence and severity of the intrinsic cardiac defects and associated congenital anomalies, rather than the type of surgical approach.
- ▶ Surgical technique is evolving, and surgical options should be considered in all cases, taking comorbidities into account.
- ▶ Antenatal ultrasonography plays an important role in detecting such anomalies which may be important for planning its further management.

Competing interests None.

Patient consent Obtained.

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