

CASE REPORT

ISOLATED ECTOPIA CORDIS: A RARE ANOMALY IN ASSOCIATION WITH CONSANGUINITY

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ABSTRACT: Thoracic type of isolated ectopia cordis is a rare anomaly with a reported prevalence of 8 per million live births. We report a case of isolated ectopia cordis in a neonate born out of second degree consanguinity, presenting with defective lower sternal wall, respiratory distress and peripheral cyanosis. He died of respiratory acidosis, dyselectrolytemia and septicemic shock before surgical intervention could be undertaken.

KEYWORDS: Ectopia cordis, sterna defects, consanguinity.

INTRODUCTION: Isolated ectopia cordis is an uncommon lethal developmental anomaly with a poor prognosis, wherein the heart protrudes through a thoraco-abdominal midline defect. It has prevalence is 5-8 per million live births with a female preponderance. Its scarcity in association with consanguinity makes it rarer. Hereby, we discuss a case of ectopia cordis with a brief review of literature.

CASE HISTORY: A live male baby weighing 2700 gm. was brought to hospital with complaints of externally beating heart and difficulty in breathing. It was born at 33 weeks of gestation to a 25 year old primigravida by preterm normal vaginal delivery. It was referred to our hospital for further management after 2 hours of birth. There was no history of infections, exposure to radiation or intake of drugs/alcohol during the period of gestation. History of second degree consanguinity was present. Antenatal scan done at 32 weeks of gestation showed a live fetus with heart beating outside the thoracic cavity. Examination revealed hypothermia, peripheral cyanosis with heart rate of 145 beats/min and respiratory rate of 62 cycles/min. A thoracic wall deformity over the middle and lower sternum was observed. The defect measured 5 cm. in length, 4 cm. in width and 2 cm. in depth. The ectopic contractile heart, entirely protruded through the sternal defect. The heart lacked pericardium, and was covered by serous membrane. The apex of the heart pointed anteriorly. No great vessels could be visualized. The face, cord, genitalia and limbs were normal. There were no other obvious anomalies. Parents were updated and they requested for comfort care. Measures were taken to prevent dehydration, electrolyte imbalance and hypothermia. Broad spectrum antibiotics were initiated. The baby died before any investigation or emergency surgical intervention could be undertaken. Autopsy was denied by relatives due to religious beliefs.

DISCUSSION: Evisceration of heart outside thoracic cavity is called ectopia cordis. The heart may be partly or completely outside. The term ectopia cordis was first described by Haller in 1706. Ectopia cordis is complete if the heart is naked, without pericardium and skin covering it.

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Partial ectopia cordis has pericardium that lies underneath the sternal surface or skin overlying the sternum. Weese (1818) and Todd (1836) described types of ectopia cordis based on position of protruding heart and extent of body wall defect as cervical, where heart lies in neck and the sternum is intact; cervico-thoracic, where the upper sternum is split and heart lies at junction of neck and thorax; thoracic, where heart protrudes through split or absent lower sternum; thoraco-abdominal, which is a distinct syndrome complex of Cantrell's pentology; lastly, abdominal where the heart passes through diaphragmatic cleft. Our case is a thoracic type with absent lower sternum, extra-thoracic visible heart without parietal pericardial covering. Defective embryologic development and differentiation of mesodermal transverse septum causes pericardial and diaphragmatic defects, while abnormal development of epi-myocardium leads to intra-cardiac anomalies. Similarly, faulty migration and fusion of paired primordial structures form ventral sternal and thoraco-abdominal wall defects.^[1] Another theory attempting to explain the pathogenesis of this anomaly is amniotic band theory, which explains development of fibrous bands owing to early disruption of yolk sac. These fibrous bands amputate fetal structures and mechanically obstruct the descent of viscera.^[2] Ectopia cordis may be isolated or in association with Cantrell's pentology. The pentad consists of a midline supra-umbilical abdominal wall defect, defect of the lower sternum, absence of the anterior diaphragm, defective diaphragmatic pericardium, and congenital intra-cardiac anomalies. Our case has an isolated form of ectopia cordis.

Most common intra-cardiac anomalies linked to ectopia cordis are inter-septal defect, pulmonary valve stenosis, Tetralogy of Fallot and left ventricular diverticulum.^[1] It may be accompanied by extra-cardiac defects like facial and cranial malformations, cleft lip, cleft palate, pulmonary hypoplasia, gastrointestinal tract defects and genitourinary system anomalies.

Although most cases are sporadic origin, Beck et al put forth association with chromosomal abnormalities of trisomy 13, trisomy 18, triploidy, etc.^[3,4] X linked recessive inheritance is explainable, since the present case has consanguinity. Martin et al attempts to explain the possible association of ectopia cordis with gene linked to Xq25-26.1 region.^[5,6]

Ultrasound scan is the best for prenatal assessment. Most cases can be diagnosed within 14-18 weeks of gestation by ultrasound scan, but few cases may be missed due to oligohydromnios.^[7] The earliest prenatal diagnosis of Pentology of Cantrell is found to be at 9 weeks.^[4] It should be supplemented with prenatal Echocardiography and Magnetic Resonance Imaging to detect intra-cardiac anomalies.

Ectopia Cordis and associated anomalies should be diagnosed in Antenatal period and steps should be taken for atraumatic delivery by caesarean section. A multidisciplinary medical team consisting of gynecologist, neonatologist, pediatric cardiologist, and pediatric surgeon should choose the best approach to this rare and severe disorder. Immediate covering of the heart and exposed abdominal contents using silastic prosthesis is recommended. The repair of omphalocele should not be delayed. Repair of the sternal, pericardial, diaphragmatic defects can be attempted at the same time. Sternal defects should be corrected in the neonatal period as the chest wall is more flexible and allows an easy approximation of sternal halves. Primary repair is preferable over sliding chondrotomies, repair by autologous grafts and prosthetic material.

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Hornberger et al performed surgical correction in 13 cases of thoracic and thoraco-abdominal Ectopia cordis and concluded that in the absence of extra-cardiac anomalies, infants with thoracic and thoraco-abdominal ectopia cordis could survive beyond early infancy and undergo successful cardiac repair.^[8] Van hoorne et al explained the prognosis depending on various clinical types of ectopia cordis and associated extra-cardiac defects. Thoraco-abdominal ectopia cordis carries better prognosis in comparison to cervical and thoracic types. Prognosis is poorer in complete form of pentalogy of Cantrell and when associated extra-cardiac anomalies. Intra-cardiac defects do not seem to be a prognostic factor.^[9]

Our case, an isolated thoracic type of ectopia cordis with second degree consanguinity, presented with fatal outcomes. In a developing country like ours, efforts towards effective prenatal diagnosis and rigorous surgical intervention should be established.

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Figure 1



Figure 2

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