

Complete Thoracic Ectopia Cordis with Craniofacial Defects & Amniotic Band – A Case Report

***Dr. Shashank Shekhar**

***Corresponding author:**

Dr. Shashank Shekhar, 401/ 4, Residential Complex, AIIMS Jodhpur, Rajasthan

E-mail: longshanks28@gmail.com

Abstract: *Ectopia cordis is a very rare anomaly defined as presence of heart partially or completely outside the thoracic cavity. A case is being reported of complete ectopia cordis thoracalis as an isolated ventral midline defect with craniofacial anomalies diagnosed prenatally using two-dimensional ultrasound at 30 weeks of gestation. Examination confirmed the ultrasound findings. The aim of the study is to report the prenatal diagnostic features and management of ectopia cordis.*

Keywords: *Ectopia cordis, extrathoracic heart, ventral midline defect.*

Introduction

Cordis ectopia is a very rare but visually striking anomaly with a reported incidence of 5.5 to 7.9 per 1 million live births¹. Despite being first noted 5000 years back, the exact etiology is still an enigma. The aim of this study is to report prenatal diagnostic features and management of ectopia cordis.

Case Report

A 24 year old primigravida at 30 weeks of pregnancy by dates reported for antenatal check-up. She has had her routine antenatal examination and investigations done except for an anomaly scan. Two dimensional ultrasound scan revealed a single live intrauterine fetus with a pulsating mass outside a small thoracic cavity in a pool of amniotic fluid, clearly recognizable as heart. No intracardiac anomaly could be found and the cardiac outflow tracts appeared normal and were seen entering the thoracic cavity from ectopically placed heart. There was large occipital encephalocele with protrusion of almost entire brain through the defect. Bilateral cleft lip was recognised. No other ventral midline defect or any other anomaly was recognised. Biometric measurements of fetus were consistent with gestational age of 27 weeks. The umbilical cord and placenta were unremarkable. The patient denied any exposure to drugs, toxins and ionizing radiations. There was no history of febrile illness. Family history for congenital malformations and genetic abnormalities was negative. Genetic amniocentesis was offered but was declined by the patient. Neonatology and pediatric surgeon's opinion was sought and patient was counseled regarding poor prognosis for this fetus and a non-aggressive management plan was agreed upon, owing to advanced gestation beyond period of viability.

Patient reported in spontaneous labour at two days past her expected date of delivery by dates and the fetus presented by breech. She delivered a severely asphyxiated live male child vaginally, weighing 2000gms. Baby died within few minutes of delivery. Detailed examination revealed a complete thoracic Ectopia cordis with no pericardial covering and cephalad orientation of the apex (Figure A). There was no other ventral midline defect. There was a large occipital encephalocele with protrusion of entire brain through this defect. A band of amniotic membrane was seen adherent to the encephalocele. Bilateral cleft lip with cleft palate was present. No other congenital malformation was found in the fetus. Autopsy was refused by the parents.

Discussion

Ectopia cordis is defined as a heart which is completely or partially placed outside the thoracic cavity. Primarily it represents a pericardial defect with eventual displacement; complete or partial, of heart outside the thoracic cavity. Depending upon the site of ectopic heart four variants have been described; cervical, thoracic, thoraco-abdominal and abdominal (2,3). Thoracic ectopia cordis is classic type (index case) and is characterized by a sternal defect allowing protrusion of heart out of chest and it's cephalad orientation. Cervical type represents retention of heart in its embryonic position in the neck, is least common and is not considered compatible with life. The thoraco-abdominal type is commonly associated with constellation of ventral midline defects termed Cantrell pentalogy; hallmark of which is ectopia cordis with omphalocele. Ectopia cordis may occur in isolation or more commonly with a host of ventral body wall defects affecting thorax, abdomen or both.

Ectopia cordis is frequently associated with a number of complex and extensive anomalies involving cranium, face, neural tube defect, intracardiac, gastrointestinal and pulmonary hypoplasia to name a few. Out of all neural tube defects, association of encephalocele seems to be most frequent with ectopia cordis(4). Even after 5000 years since its first description the exact cause of ectopia cordis still evades the researchers. Out of all major theories put forth, the body wall fusion failure hypothesis is the principal leading theory. Lateral body folds form during third post fertilization week as a combination of parietal layer of lateral plate mesoderm and overlying ectoderm. They move ventrally to meet in midline and the closure occurs by the end of fourth week of gestation(5). The closure of ventral body wall has been likened to neural tube closure for two reasons. First the cellular and molecular processes involved are similar and secondly they both share a similar embryonic time line of development, so that most neural tube and ventral body wall defects have their origin during fourth week of development. Unlike ventral midline defect (VMD) and neural tube defects(NTD), cleft lip or cleft palate are separated widely on embryonic time line as latter are determined around 5th to 6th week of development. However VMD and facial defects are spatially similar in that they both occur on the ventral aspect of the embryonic midline (4).

Because of rarity of the disease, the management had been slow to evolve. Not so long ago the diagnosis of ectopia cordis was made in delivery room but with the wide use and improvement of ultrasound equipment and increasing awareness of ectopia cordis, diagnosis as early as 10weeks have been reported(6). Once diagnosed antenatally a vigorous approach should be adopted to search for associated anomalies. This helps in accurate prognostication and classification of the anomaly. Though considered to be of sporadic occurrence reports of associated chromosomal anomalies like trisomy 18 and Turner syndrome have emerged in literature, hence karyotyping should be offered to all as it helps in improving genetic counseling(7). Though ectopia cordis is nearly fatal in all cases, those with minimal or no associated anomalies may stand a chance of survival after surgical correction. Hence some authors suggest a planned atraumatic abdominal delivery in such cases followed by surgical correction (8). However pregnancy termination before viability is advisable in view of frequent association with multiorgan anomalies, limited clinical experience with such a rare condition and near dismal surgical results (9, 10). In case of a third trimester diagnosis a non-aggressive approach should be adopted. Majority of cases of ectopia cordis are stillborn. Vaginal delivery poses a major risk due to prolonged cardiac compression and likely rupture of atrial diverticulum and omphalocele sac.

Conclusion

Ectopia cordis carries a very poor prognosis nonetheless it is easily recognisable on ultrasound prenatally. Ectopia should be precisely localized and classified accurately. A detailed search for associated anomalies should be made. Karyotyping should be offered as part of obstetrical management. Termination of pregnancy is the mainstay of management if diagnosed before viability however a non-aggressive approach in the third trimester should be considered.

Conflict of Interests: None

Fig 1 Frontal view of the fetus showing completely bare heart outside thoracic cavity with cephalad orientation of the apex. Note the big encephalocele and attached amniotic membrane. Umbilical cord insertion on the abdominal wall is normal and there is no abdominal wall defect.



Fig. 1:

References

1. Hornberger LK, Colan SD, Lock JE, Wessel DL, Mayer JEI Outcome of patients with ectopia cordis and significant intracardiac defects. *Circulation* 1996; 94:32-7.
2. Van Praagh R, Weinberg PM, Smith SD, Foran RB. In: Adams FA Emmanoulides GC, Reimensneider TA, eds. *Heart disease in infants, children and adolescents*. 4th ed. Baltimore: Williams and Wilking.1989:570
3. Leca F, Thibert N, Khoury W, Fermont L, Laborde F, Dumez Y. Extrathoracic Heart (ectopia cordis): report of two cases and review of literature. *Int J Cardiol* 1989; 22:221-8.

4. Carmi R, Boughman JA. Pentalogy of Cantrell and associated midline anomalies: a possible midline developmental defect. *Am J Med Genet* 1992; 42:90?
5. Sadler TW. The embryological origin of ventral body wall defects. *Semi Pediatr Surg* 2010 Aug; 19(3):209-14.
6. Liang RI, Huang SE, Chang FM. Prenatal diagnosis of ectopia cordis at 10 weeks of Gestation using two-dimensional and three-dimensional ultrasonography. *Ultrasound Obstet Gynecol* 10(1997)137-139.
7. Klingensmith WCIII, Cioffi-Ragan DT, Harvey DE. Diagnosis of ectopia cordis in the Second trimester. *J Clin Ultrasound* 1988; 16:204-6.
8. Diaz JH. Perioperative management of neonatal ectopia cordis: report of three cases. *Anest Analg* 1992; 75:833-7.
9. Morales JM, Patel SG, Duffy JA, Villareal RL, Simpson JW. Ectopia cordis and other Midline defects. *Ann Thorac Surg* 200; 70:111-4. (Dismal surgery results)
10. Chopra S, Kalpdev A, Suri V, Aggarwal N, Rohilla M. Complete ectopia cordis with Anencephaly: a case report. *J Clin ultrasound* 2010 May; 38(4):212-4.