Indian Journal of Obstetrics and Gynecology Research 2021;8(1):127–129

Content available at: https://www.ipinnovative.com/open-access-journals

Indian Journal of Obstetrics and Gynecology Research

Journal homepage: www.ijogr.org

Case Report

Ectopia cordis with omphalocele and exencephaly – A case report

Surabhi Derkar[1,*]

[1] *Dept. of Radiodiagnosis, Shree Sai Diagnostic Centre, Nagpur, Maharashtra, India*

ARTICLE INFO

Article history:
Received 07-11-2020
Accepted 23-11-2020
Available online 13-03-2021

Keywords:
Ectopia cordis
Omphalocele
Exencephaly
Pentalogy of Cantrell

ABSTRACT

Ectopia cordis is an extremely rare congenital anomaly where the heart is in extrathoracic location partially or completely. We report a case of a 25-year-old primigravida with uneventful pregnancy, no relevant family or personal history, in which the prenatal fetal ultrasound performed at 19 weeks of gestation revealed an anterior abdominal wall defect with exteriorisation of the heart and exencephaly. The pregnancy was terminated after counselling parents with baby weighing 119 gm. Sonographic findings were confirmed on post-natal examination.

1. Introduction

Ectopia cordis is a rare disease that is defined by the abnormal position of the heart outside the thoracic cavity associated with defects in the abdominal wall, diaphragm, sternum and intracardiac malformations.

The cause of this pathology is currently unknown with poor prognosis.

It may appear as an isolated anomaly or in association with other malformations like omphalocele, congenital heart disease or pentalogy of Cantrell syndrome.

Byron classified Ectopia cordis into four types: cervical, thoraco-cervical/thoracic, thoraco-abdominal and abdominal.[1]

Cervical (5%): The heart is in its embryonic position in cervical region with intact sternum.

Thoraco-cervical/thoracic (65%): The heart lies outside the thorax, partially or completely with sternal defect.

Thoraco-abdominal (20%): It is commonly associated with constellation of ventral midline defects termed pentalogy of Cantrell; hallmark of which is ectopia cordis with omphalocele.

Abdominal (10%): The heart is in abdominal cavity entailing a defect in the diaphragm.

Here we report a case of a variant of Cantrell's syndrome presenting with complete ectopia cordis (thoraco-abdominal), omphalocele and exencephaly.

2. Case Report

A routine prenatal ultrasound examination in a 25-year-old primigravida was performed at 19 weeks of gestation for the evaluation of fetal well-being. Transabdominal sonographic evaluation using a Toshiba (Nemio XG) equipped with 3.75 MHz convex transducer demonstrated a live fetus with parameters corresponding to 15 weeks gestation, with exencephaly, anterior abdominal wall defect including herniated liver and ectopia cordis with a large omphalocele containing the intestines, in cephalic presentation.

The mother was referred to a tertiary setting following the detection of multiple fetal anomalies. Her medical and obstetric histories were unremarkable. Parents were non-consanguineous. There was no family history of congenital anomalies, genetic abnormalities, or history related to ectopia cordis. Parents were counseled regarding poor prognosis of the condition with complex presentation and multi- staged corrective surgical procedures and therefore

* *Corresponding author.*
E-mail address: surabhiderkar1992@gmail.com (S. Derkar).

https://doi.org/10.18231/j.ijogr.2021.026

they opted for pregnancy termination.

Post-natal examination revealed an anterior thoraco-abdominal wall defect, ectopic contractile heart (Figure 1), an edematous liver and spleen with loops of small intestine and colon outside the abdominal cavity covered by a membrane with umbilical cord directly inserting into the herniated segment (Figure 2). The face was dysmorphic with absent cranial bone and bulging orbits along with complete herniation of brain parenchymal tissue (Figure 3). The thoracic spine was showing kyphosis. All the four limbs were normal. The baby weighed 119 gm at 19 weeks of gestation. Parents were updated post-surgery, however they did not consent for autopsy.

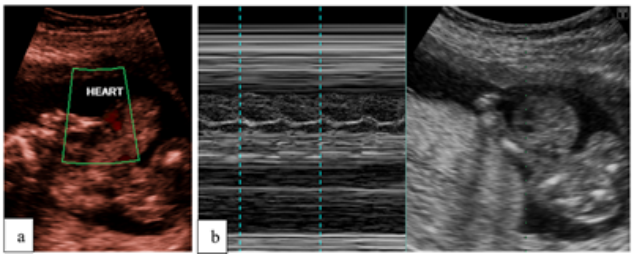

Fig. 1: Ectopic contractile heart

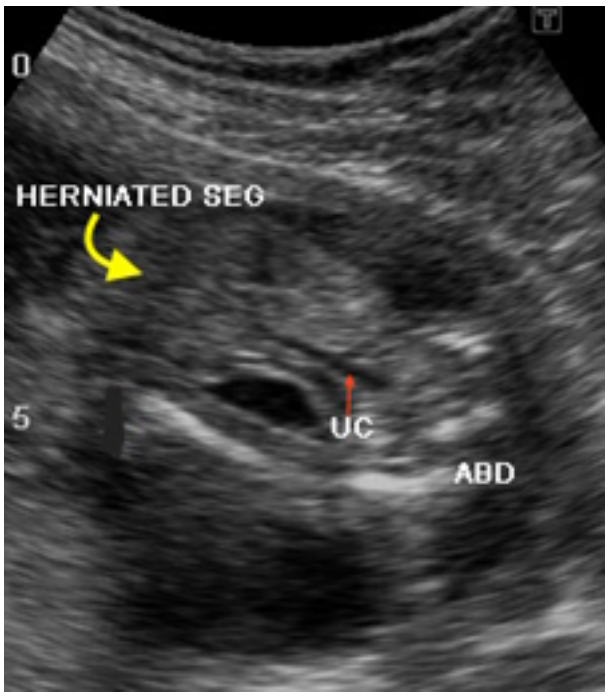

Fig. 2: Omphalocele

3. Discussion

Ectopia cordis (EC) is a rare congenital malformation in which the fetal heart partially or completely lies outside the thoracic cavity with reported incidence of 5 to 8 per million live births. Abott first coined the term Ectopia cordis in 1998 with first prenatal diagnosis reported by Wicks et al. in 1981 at 34 weeks of gestation.[2] The prenatal confirmation of EC with severe congenital heart disease can be done by vaginal echocardiography at 10-12 weeks of gestation or by abdominal echocardiography by 20-22 weeks.

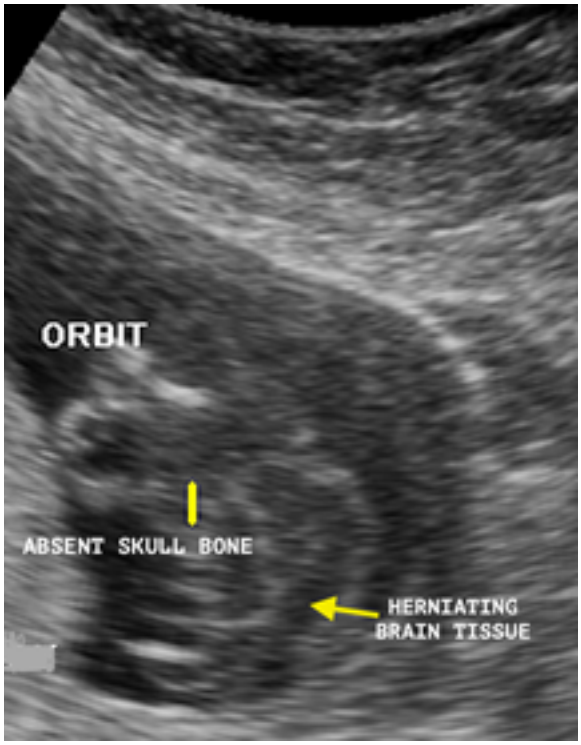

Fig. 3: Exencephaly

Complete or incomplete failure of midline fusion at 9^{th} embryonic week results in disorders varying from isolated EC to complete ventral evisceration. Although several theories have been proposed, genesis of EC has not been fully explained. Rupture of the chorion and/or yolk sac at approximately three weeks gestation is postulated to interfere with normal cardiac descent. Other popular theories include mechanical teratogenesis and amniotic band syndrome.[3,4]

Omphalocele is the most commonly associated anomaly. Since omphaloceles are also assumed to be secondary to anterior fusion defects, the association of an omphalocele with ectopia cordis as in the present case is more likely.[5] In our case, which was diagnosed at 19 weeks of gestation, there is a midline abdominal wall defect, through which bowel loops with liver and spleen are herniating, covered by a peritoneal membrane. The umbilical cord insertion is seen directly into the omphalocele.

Ectopic heart is one of the distinctive anomalies seen in patients presenting with the rare syndrome of pentalogy of Cantrell which comprises midline supraumbilical

abdominal wall defect, deficiency of the anterior diaphragm, defect of the lower sternum, defect in diaphragmatic pericardium, and congenital heart disease. Ectopia cordis cases associated with the Pentalogy of Cantrell reported in the literature had central nervous system and craniofacial malformations in 8 cases and only 3 of those cases had exencephaly.[6–8] Pentalogy of Cantrell associated with exencephaly has been reported only in 3 cases and association with craniorachischisis has been reported in only 2 cases.[9] In our case, absent cranial vault is noted with herniated brain tissue dangling in the amniotic fluid.

The combination of three-dimensional ultrasound with Doppler allows for a more precise early diagnosis. Most cases of EC are diagnosed in the second trimester which allows for the adequate delineation of the associated anomalies present in almost 90% of the cases and hence enables the parents to take informed and clear decisions concerning the fate of the pregnancy.

Although ectopia cordis is generally considered an isolated, sporadic malformation, its occasional association with chromosomal abnormalities suggests that prenatal karyotyping should be considered in the management of these cases, especially for thoraco-abdominal ectopia cordis. The core issue with this disorder is related to the lack of the normal protection of the heart by the sternum rendering it at increased risk from direct trauma. In addition, these patients are susceptible to recurrent chest infections due to the paradoxical movement of the lungs.

The prognosis, although poor, depends on the degree of the intra-cardiac and associated malformations, the variability of ectopia cordis and therefore the technical equipment for neonatal resuscitation.[10] The survival rate is very low even after surgical correction in selected cases. Most parents choose termination of the pregnancy, and for this reason an early diagnosis becomes very desirable since this has repercussions on the method of termination and its safety profile.

4. Conclusion

Complete ectopia cordis is uniformly contemplated to be fatal, however with poor prognosis. Ultrasonography provides a significant role in the assessment of ectopia with its precise location and classification in view of the different prognosis. Magnetic resonance imaging is also becoming conventional in prenatal assessment to document and plan for management of complicated congenital anomalies.

Death usually occurs within the 1st few days of life as a result of infection, cardiac failure, or hypoxemia, hence its management needs prompt medical care and surgical intervention taking co-morbidities under consideration. In light of limited data, association of multiple anatomic defects, and limited clinical experience with such a rare condition, termination of pregnancy is advisable. Therefore, our patient was counseled in detail about the malformation, and termination of pregnancy was carried out.

5. Source of Funding

None.

6. Conflict of Interest

The authors declare that there is no conflict of interest.

References

1. Byron F. Ectopia cordis: report of a case with attempted operative corrections. *J Thoracic Surg*. 1949;17:717–22.
2. Wicks JD, Levine MD, Mettler FA. Intrauterine sonography of thoracic ectopia cordis. *Am J Roentgenol*. 1981;137(3):619–21. doi:10.2214/ajr.137.3.619.
3. Kaplan LC, Matsuoka R, Gilbert EF, Opitz JM, Kurnit DM, Reynolds JF. Ectopia cordis and cleft sternum: Evidence for mechanical teratogenesis following rupture of the chorion or yolk sac. *Am J Med Genet*. 1985;21(1):187–99. doi:10.1002/ajmg.1320210128.
4. Bieber FR, Mostoufi-zadeh M, Birnholz JC, Driscoll SG. Amniotic band sequence associated with ectopia cordis in one twin. *J Pediatrics*. 1984;105(5):817–9. doi:10.1016/s0022-3476(84)80314-5.
5. Hauge M, Bugge M, Nielsen J. Early prenatal diagnosis of omphalocele constitutes indication for amniocentesis. *Lancet*. 1983;322(8348):507. doi:10.1016/s0140-6736(83)90528-7.
6. Bognoni V, Quartuccio A, Quartuccio A. First-trimester sonographic diagnosis of Cantrell's pentalogy with exencephaly. *J Clin Ultrasound*. 1999;27(5):276–8. doi:10.1002/(sici)1097-0096(199906)27:5<276::aid-jcu7>3.0.co;2-s.
7. Denath FM, Romano W, Solcz M, Donnelly D. Ultrasonographic findings of exencephaly in pentalogy of cantrell: Case report and review of the literature. *J Clin Ultrasound*. 1994;22(5):351–4. doi:10.1002/jcu.1870220512.
8. Peer D, Moroder W, Delucca A. Prenatal diagnosis of the pentalogy of Cantrell combined with exencephaly and amniotic band syndrome. *Ultraschall Med*. 1993;14:94–94.
9. Polat I, Gül A, Aslan H, Cebeci A, Ozseker B, Caglar B. Prenatal diagnosis of pentalogy of Cantrell in three cases, two with craniorachischisis. *J Clin Ultrasound*. 2005;33:308–11.
10. Taksande AM, Vilhekar KY. A case report of ectopia cordis and omphalocele. *Indian J Hum Genet*. 2013;19(4):491–3. doi:10.4103/0971-6866.124384.

Author biography

Surabhi Derkar, Consultant Radiologist

Cite this article: Derkar S. Ectopia cordis with omphalocele and exencephaly – A case report. *Indian J Obstet Gynecol Res* 2021;8(1):127-129.