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Pathology

PENTALOGY OF CANTRELL-THE RAREST AMONGST THE RARE

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ABSTRACT Pentalogy of Cantrell(PC) is a rare congenital anamoly that causes defects involving the heart, pericardium, diaphragm, abdominal wall and lower sternum.

This case report presents a female fetus, who was delivered at 20 weeks of conception and weighed 280 g. She had the five anatomical defects known as Complete Cantrell's Pentalogy. Most infants do not develop all of the potential defects, which may be referred to as incomplete pentalogy of Cantrell. The variability of the disorder from one individual to another can be significant.PC is a life threatening congenital anomaly and a recognisable cause of fetal and neonatal morbidity due to cardiac insufficiency and anatomical defects.An early diagnosis with increased awareness of the disease is a crucial factor for a timely approach to manage the critically ill infant and to offer potential step-wise corrective surgeries for a favourable outcome and substantial reduction in morbidity.

KEYWORDS : Diaphragmatic defects, Ectopia Cordis, Omphalocele, Pentalogy of Cantrell, Thoraco-abdominal wall defect

INTRODUCTION

PC is a rare pathology in fetuses which require intensive neonatal care and is asoociated with high mortality and morbidity.

Cantrell in 1958 first described the full spectrum of PC.[1] The pentad of PC comprises of -1) Ectopia cordis and intracardiac anomalies; 2) lower sternal defect; 3) midline supraumbilical thoraco-abdominal wall defect; 4) anterior diaphragmatic defect; and 5) defect of diaphragmatic part of pericardium that results in relation between pericardial cavity and peritoneum.[1]

Few case reports in literature have been classified as full spectrum, which have all of the five defects. Incomplete forms with lesser defects of the pentalogy of Cantrell have also been mentioned.[2,3,4] The prognosis in these cases depends on the severity of the anomalies. There are a few reported survivors after corrective surgery. [2]

The diagnosis of PC can often be made prenatally with a fetal ultrasound.To evaluate, the extent of the involvement of the heart,an echocardiography can be performed.The exact cause of PC is not known. Most of these cases occur sporadically.

CASE REPORT

A 25 year old primi reported to us for a routine anamoly scan at 20 weeks of gestation. Antenatal ultrasound revealed ectopia cordis and midline supraumbilical defect of thoraco-abdominal wall with evisceration of liver and bowel loops(Fig 1). Her medical and obstetric history were unremarkable. The marriage was non-consaguineous. Parents were counscelled and the pregnancy was terminated. At birth the fetus ectopic heart was faintly contractile for less than a minute. The fetus was received by the Department of Pathology for autopsy. Weight of the fetus was 280gms. Crown rump and crown heel length was 14.7 and 27 cms respectively. The head circumference,chest circumference and abdominal girth was 18, 15.5 and 11.5cms respectively. External examination revealed a large anterior thoraco-abdominal defect extending from the upper third of sternum till supra umbilical level ,ectopic heart, liver and oedematous bowel loops outside the abdominal cavity(Fig 2). Lower half of the body of sternum and the xiphisternum were absent. No facial dysmorphism was noted.All four limbs were within normal limit.

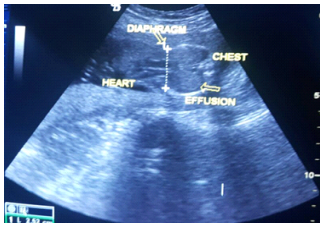


Fig1-Fetal ultrasound scan showing ectopia cordis with bilateral pleural effusion.



Fig 2-Fetus at birth with Pentalogy of Cantrell

DISCUSSION

PC affects 5.5 million per live births.[2] There is slight male to female preponderance 2.7:1. This condition is believed to be caused by failure in embryogenesis that occurs during the first weeks after conception. As Cantrell suggested , it is a failure of embryologic development of a segment of the lateral mesoderm occurring at 14–18 days after conception .[1] Consequently, the transverse septum of the diaphragm does not develop and the paired mesodermal folds of the upper abdomen do not migrate ventromedially.[3] The frequency rate of occurrence of complete spectrum of PC is much lower than the incomplete spectrum of PC.[2] A classification based on the number of anomalies in PC was proposed by Toyoma.The classification is related to association between complexity of intra-cardiac anomalies and increased mortality.[4] On the basis of anatomical location,Ectopia Cordis can be classified as cervical,cervicothoracic,thoracic and thoracoabdominal types.[5] Ventricular septal defects are present in 100% of cases, atrial septal defects (53%), tetralogy of Fallot (20%), and a left ventricular diverticulum in 20% .[1] Many case reports have established the association of PC with other congenital defects like cleft lip/ palate, encephalocele, hydrocephalus, craniorachischisis, absence of radius or tibia and club feet.[6,7]

PC can be corrected by multistage corrective cardiovascular surgeries which involves initial closure of the thoraco-abdominal wall defects.The degree of Ectopia Cordis, size of the omphalocele,and the complexities of the cardiac anamolies decides the strategy of the surgeries performed.Due to severe intracardiac defects,most of the cases unfortunately result in termination of pregnancy,postpartum fetal death or early neonatal death.

CONCLUSION

Increasing awareness about this rare congenital anamoly amongst the ultrasonologist will increase chances of detecting it before viability which gives the parents a wider range of options for surgical intervention or termination.

REFERENCES

1. Cantrell JR, Haller JA, Ravitch MM. A syndrome of congenital defects involving the abdominal wall, sternum, diaphragm, pericardium, and heart. Surg Gynecol Obstet. 1958;107:602–14.
2. J.H.L. van Hoorn, R.M.J. Moonen, C.J.R. Huysentruyt, L.W.E. van Heurn, J.P.M. Offermans, A.L.M.T. MulderPentalogy of Cantrell: two patients and a review to determine prognostic factors for optimal approach.Eur J Pediatr.2008; 167 : 29-35
3. H. Takayasu, Y. Kitano, T. Kuroda, N. Morikawa, H. Tanaka, A. Fujina, et al.Successful management of a large fetal mediastinal teratoma complicated by hydrops fetalis.J Pediatr Surg. 2010;21-24
4. W.D. Toyama.Combined congenital defects of the anterior abdominal wall, sternum, diaphragm, pericardium, and heart: a case report and review of the syndrome.Pediatrics.1972: 50 :778-792
5. J.M. Morales, S.G. Patel, J.A. Duff, R.L. Villareal, J.W. SimpsonEctopia cordis and other midline defects.Ann Thorac Surg.2000;70:111-114
6. S. Hirokawa, H. Uotani, H. Okami, K. Tsukada, T. Futatani, I. HashimotoA case of congenital midline cervical cleft with congenital heart disease J Pediatr Surg.2003;38:1099-1101
7. R. Smigiel, A. Jakubiak, M.P. Lombardi, W. Jaworski, R. Slezak, D. Patkowski, et al.Co-occurrence of severe Goltz-Gorlin syndrome and pentalogy of Cantrell – case report and review of the literature Am J Med Genet A.2011;55 : 1102-1105