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Case Report

A rare case of incomplete Pentalogy of Cantrell diagnosed on antenatal ultrasound

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ABSTRACT

Pentalogy of Cantrell (POC) is a rare and complex congenital anomaly characterized by midline defects involving the abdominal wall, sternum, diaphragm, pericardium, and heart. The condition is often fatal and poses significant diagnostic and management challenges. We report a rare case of incomplete POC diagnosed during a routine second-trimester anomaly scan in a primigravida. The pregnancy was medically terminated after thorough counseling. This case highlights the importance of early prenatal diagnosis, multidisciplinary evaluation, and the ethical considerations involved in the management of complex fetal anomalies.

Keywords: Pentalogy of Cantrell, Midline defect, Primigravida

INTRODUCTION

Pentology of Cantrell (POC) is a rare, congenital disorder, originally defined by Cantrell and colleagues in 1958 and later revised by Toyama in 1972.[1] The pentad involves the complete or partial expression of five congenital birth defects, originating at the lower sternum, anterior diaphragm, diaphragmatic pericardium, midline /ventral supraumbilical abdominal wall region, and heart. Cantrell and associates criteria included the absolute presence of all five defects. Toyama subsequently published a modified classification for POC, enabling inclusion of patients who present with variable quantities of the classic spectrum of defects.[2] The incidence of POC is 1 in 5.5 million live births.[3] Toward the end of the 20th century, the mortality rate for this rare disorder was estimated at 52%. In 2014, Zhang and colleagues reported a mortality rate of 61%. The increased mortality may be attributed to Toyama's wider diagnostic classification, lower survivor rates, or selection bias, which may be influenced by the limited number of reported cases.[4]

CASE REPORT

A 24-year-old primigravida at 19 weeks 4 days of gestation, presented to antenatal outpatient department of tertiary healthcare hospital for routine antenatal checkup and was diagnosed with incomplete fetal POC in level 2 scan. On Ultrasonography intra uterine pregnancy with gestation age of 18 weeks 2 days +/- 2 weeks with heart rate of 136 bpm with defect in the anterior abdomen and thoracic wall. The abdominal wall defect is paramedian with umbilical cord insertion seen separately. Herniation of abdominal organs (liver, gall bladder, intestine) noted which were free floating with no membrane covering it. Associated herniation of heart is also noted adjacent to the above herniated abdominal organs. (Figure 1 and 2).

Detailed study of the heart could not be done due to limited evaluation/ectopic position.

Patient and attendants were counselled regarding the prognosis and need for termination of pregnancy. An anomalous female fetus weighing 165 gm was aborted completely with placenta after medical termination of pregnancy.

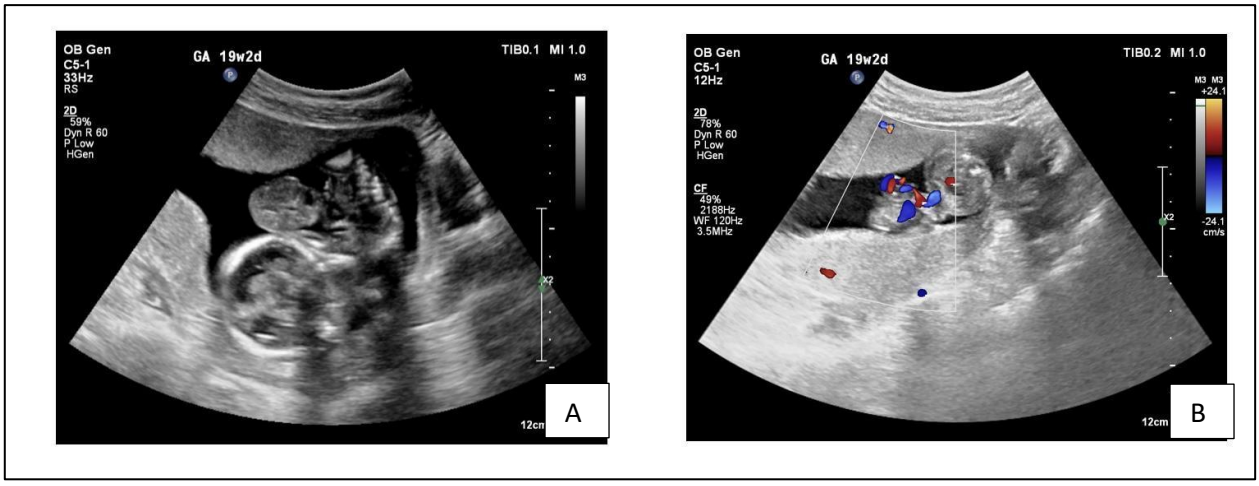

Figure 1 (A and B): Sonographic representation of a 19 weeks 2 days fetus with omphalocoele.

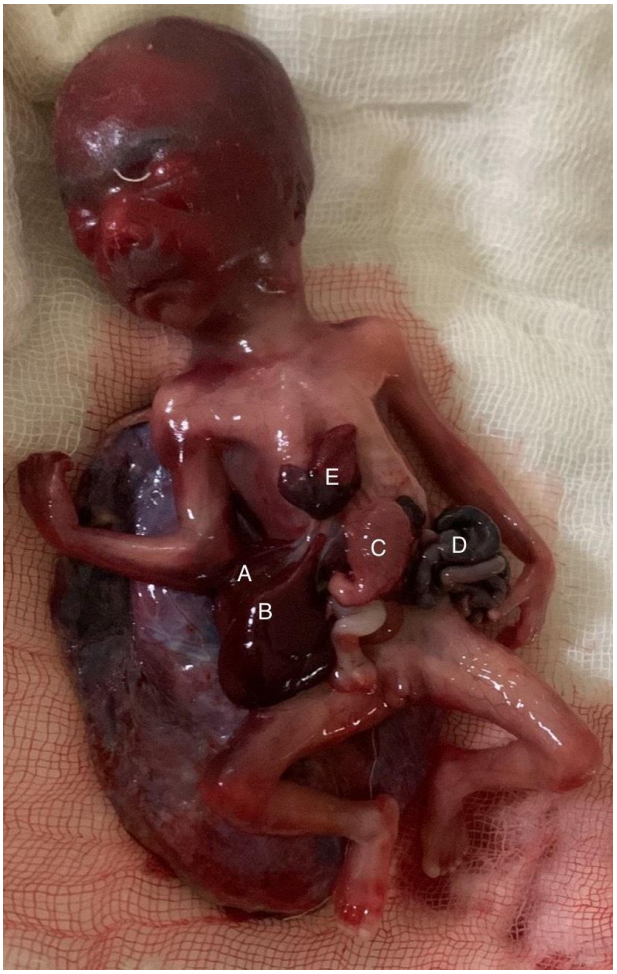

Figure 2: 19 weeks abortus with pentalogy of cantrell - omphalocoele (A) pancreas, (B) liver, (C) gastroschisis, (D) intestine and (E) ectopia cordis.

Various anomalies noted in female fetus:

Nuchal fold thickened, cystic hygroma present

Large anterior abdominal wall defect seen: gastroschisis - with liver, pancreas stomach intestines seen completely outside abdominal wall.

Normal palmar crease.

Complete absence of xiphoid process

Sternal defect with congenital diaphragmatic defect

No neurological anomalies seen

No e/o meningomyelocele

No e/o spina bifida

A diagnosis of incomplete pentalogy of cantrell was made.

Embryology

The genesis of defective fetal development associated with POC most likely occurs within the first 8 weeks after fertilization of the zygote. During this phase, the embryo begins a complex sequence of cellular differentiation. This critical embryonic period occurs during the second to eighth week of gestation and encompasses both blastogenesis and organogenesis. During this timeframe, the embryonic sac, located at the merger of the amniotic cavity and yolk–gut cavity, differentiates into three germinal tissues, the ectoderm (top layer), mesoderm (middle layer), and endoderm (inner layer). “Folding” or rolling of these three flats, stacked layers of cells occurs

during the third to fifth weeks of gestation, forming a tube-like shape with the ectoderm encapsulating the entire structure. The germinal tissues then begin to differentiate to facilitate organogenesis and the development of other essential structures. The ectoderm differentiates into the surface ectoderm and neuroectoderm, which induces the formation of the future epidermis and neural tube. The mesoderm differentiates into the paraxial mesoderm, intermediate mesoderm, and lateral plate mesoderm, which will develop into the peritoneal and pleural membranes as well as the bones, cartilage, tissues, muscles, vessels, reproductive, excretory, and urogenital systems. Finally, the endoderm differentiates in a cephalocaudal direction to form the endocrine glands, respiratory system, and gut. Failure during any phase of the embryonic period may result in fetal maldevelopment and expression of congenital defects consistent.

Management and treatment strategies

The initial management and stabilization of infants with POC are critical hence, management in tertiary care hospital with appropriate NICU facility. The immediate time frame after delivery must be used to inspect, assess, diagnose, and determine whether surgical correction of associated defects is feasible. Initial management strategies include neonatal resuscitation with stabilization, the extent of which is closely aligned with parental wishes and prognostic indicators. Protection of the presenting defects during neonatal resuscitation is imperative.

The presence of an omphalocele or abdominal wall defect, the degree of prolapse of the heart, the size and degree of herniation of the diaphragmatic defect, and any malformations of the heart and/or great vessels all predispose the neonate to hemodynamic instability. Initial correction of metabolic and hemodynamic alterations, in conjunction with ventilatory support, may be indicated. Extracorporeal membrane oxygenation and inhaled nitric oxide, inotropes, and fluid resuscitation may also be required. Prostaglandins may also be required in the case of ductal-dependent congenital heart dis- ease to maintain dilation of the ductus arteriosus. Antibiotic therapy should be initiated for prophylaxis against sepsis, a significant risk factor associated with open sternal defects. Following the immediate resuscitation period, an assessment of the patient's candidature for surgical repair is indicated. Surgical intervention may be palliative or corrective. Primary surgical goals include correction of complex intracardiac defects, ventral hernia, and diaphragmatic defects. Determination of the timing and sequence of surgical repair of thoracoabdominal defects versus intracardiac lesions depend on the clinical presentation.

Primary closure of the omphalocele can increase thoracic pressure and respiratory or cardiovascular compromise. Therefore, avoidance of excessive compression of the heart during thoracoabdominal defect repair is indicated to reduce the risk for hemodynamic instability and/or myocardial ischemia. Perioperative and postoperative management strategies with primary closures include maintaining fluid and electrolyte balance as well as body temperature homeostasis due to exposure of the viscera.

In the case of POC with associated hepatopulmonary fusion, surgical repair is particularly complicated because of the difficulty in separating the tissues. Arrhythmias are common in the postoperative period owing to increased abdominal and thoracic pressures, which can lead to compromised venous return and cardiac output. Additional postsurgical risks include tachyarrhythmias, hypotension, rupture of the diverticulum, bradycardia, and heart failure. Other potential complications include skin infections, pulmonary dysfunction, bradycardia, hypotension, rupture of the diverticulum, pulmonary hypertension, heart failure, and cardiac arrest.

Management strategies for patients eligible for delayed surgical repair may include encouragement of epithelialization of the surrounding skin of the omphalocele with mercurial solution or benzalkonium chloride solution or with sterile gauze dressings.[5-7]

DISCUSSION

We present a rare case of POC. The etiology of this condition is not known. It is often described as being sporadic in nature. This condition involves disruption of the abdominal wall and the thoracic wall. In our case, all the features of POC were seen, including, lower sternal defect, midline supraumbilical thoracic and abdominal wall defects, diaphragmatic defect, cardiac defect, and ectopia cordis. POC can include other fetal malformations away from the midline defect. Conditions like craniofacial anomalies, cleft lip and palate, central nervous system anomalies, skeletal anomalies and clubfoot, polysplenia, and gallbladder agenesis can be seen.

The exact etiology of pentalogy of Cantrell is unknown. The general belief that the problem started in very early embryonic life, mostly likely within the first 3 weeks of the embryonic life where there is a failure of the development in the lateral mesoderm. The failure in development could be due to a number of factors which include gene mutation, chromosomal anomaly, or disrupted blood supply. In some cases of POC, there are encephalocele and facial defects such as cleft lip and palate. It is also possible in cases of POC, to have the accumulation of fluid leading to pleural effusion, this could be due to cardiac failure. Skeletal malformation is not very often seen. The diagnosis of POC can be made by ultrasound with great degree of accuracy after 12 weeks of gestation.[4-9] In the first trimester, however, physiological herniation of the fetal bowel can make the diagnosis difficult especially in the mild form of POC. A 2D ultrasound is generally adequate to make the diagnosis in this condition. 3/4D ultrasound is hardly needed in this situation, however it may help to counsel the parents and the couple may understand the 3/4D picture better. Other diagnostic modalities such as magnetic resonance imaging

(MRI) and computed tomography (CT) scan are very rarely needed. The prognosis in this condition is very poor.[5,10,11] The survival rate even after a very complex medical and surgical intervention is very poor. The mean survival rate in most cases is hours rather than days and years.

CONCLUSION

Cantrell's pentad includes defects involving the lower sternum, anterior diaphragm, diaphragmatic pericardium, midline/ventral abdominal region, and heart. A fetus presenting with any lesions associated with this pentad warrants a thorough evaluation to comprehensively assess mortality risks, the feasibility and eligibility for surgical repair in the neonatal period or beyond, and related moral and ethical implications. Incomplete expression of POC is reported in the literature; therefore, the documentation of any one or more of these defects prenatally warrants further evaluation with 2D/3D ultrasound or MRI.

Management and treatment strategies, including surgical repair, are initiated in sequence on the basis of the severity of the neonate's defects. Although not yet supported by robust evidence, a conservative approach with a staged surgical repair initiated after the immediate neonatal period may be associated with improved outcomes. Further investigation of this hypothesis is indicated. Most cases, however, are associated with ominous findings that result in the need for palliative care shortly after delivery. A multidisciplinary approach by allied health and medical experts and nursing is essential to achieve optimal outcomes for the foetus, neonate, and family unit.

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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(5):602-14.
2. Toyama WM. Combined congenital defects of the anterior abdominal wall, sternum, diaphragm, pericardium, and heart: A case report and review of the syndrome. Pediatrics. 1972;50(5):778-92.
3. Iacobacci JJ, Morrow WR, Tweddell JS, Huhta JC, Gutierrez FR. Pentalogy of Cantrell: Review of the literature and management guidelines. Pediatr Cardiol. 1994;15(4):205-10.
4. Hornberger LK, Colan SD, Lock JE, Wessel DL, Mayer JE. Outcome of patients with ectopia cordis and significant intracardiac defects. Circulation. 1996;94(9):1132-7.
5. Vazquez-Jimenez JF, Muehler EG, Daebritz SH, Uhlemann F, Meisner H. Successful surgical management of complete pentalogy of Cantrell. Ann Thorac Surg. 1998;65(2):537–9.
6. Morales JM, Patel SG, Duff JA, Villareal RL, Simpson JW. Ectopia cordis and other midline defects. Ann Thorac Surg. 2000;70(1):111-4.
7. Van Hoorn JH, Moonen RM, Huysentruyt CJ, Nijhuis JG, van Heurn LW, Mulder AL. Pentalogy of Cantrell: Two patients and a review to determine prognostic factors for optimal approach. Eur J Pediatr. 2008;167(1):29-35.
8. Zhang X, et al. Pentalogy of Cantrell: Prenatal diagnosis and postnatal outcomes. J Matern Fetal Neonatal Med. 2014.
9. Bhatia N, Mishra S, Ranjan R. Pentalogy of Cantrell: A case report with literature review. J Clin Neonatol. 2016;5(1):58–60.
10. Sharma A, et al. Pentalogy of Cantrell: A rare case diagnosed on antenatal scan. J Clin Diagn Res. 2017.
11. Harjai MM, et al. Prenatal diagnosis and management of Pentalogy of Cantrell. J Fetal Med. 2019.

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