COMPLETE PENTALOGY OF CANTRELL: CASE REPORT AND REVIEW OF LITERATURE

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Abstract

Introduction: Pentalogy of Cantrell (POC) is a rare congenital anomaly with two sub-categories: complete or partial. Complete has all five defects (anterior diaphragmatic defect, ventral abdominal wall defect, pericardial defect, intracardiac anomalies, and lower sternal defect). The first documented case was described by Cantrell et al. in 1958. Both sporadic and genetic causes have been proposed for POC, with a reported incidence of 5 - 10 cases per one million live births and various clinical presentations. The prognosis of POC depends on the severity of the defects and the associated cardiac anomalies. This case report seeks to increase awareness of this condition, emphasize the need for appropriate counselling in our environment, and review the literature on previous reported cases' outcomes.

Case Presentation: A 5-hour-old term female neonate was referred to our Children Emergency Center with multiple defects (ectopia cordis, sternal cleft, omphalocele, and limb abnormalities) with dysmorphic facies. A diagnosis of POC was made, and multidisciplinary management was instituted. The eviscerated heart and epigastric omphalocele were dressed and evaluated for palliative surgical care. However, she developed complications with sepsis and electrolyte derangements and died from multi-organ failure before any surgical intervention could be carried out.

Conclusion: Though rare, the Pentalogy of Cantrell in its classical form does occur in our environment. The presence of extracardiac and limb deformities worsens the outcome. Based on poor outcomes, there is a need to emphasize appropriate antenatal and postnatal assessments to provide effective counselling on termination, neonatal palliative care, and surgical repairs as appropriate.

Keywords: *Pentalogy of Cantrell; Birth defect; Ectopia cordis; Congenital anomaly*

Introduction

Pentalogy of Cantrell (POC) is a medical condition with five defects affecting the heart, pericardium, diaphragm, sternum, and abdominal wall. Not too many cases have been documented in the literature. The cases are mostly sporadic. Some familial cases have been reported, suggesting that genetic factors may play a role in the development of this disorder.[1] Ideally, Pentalogy of Cantrell [2] should be detected during an antenatal ultrasound scan rather than the first presentation at birth. A male predominance of 1.35 to 1 has been reported. [3] Though of multifactorial etiology, Cantrell et al. attributed it to a developmental failure of a segment of the lateral mesoderm at about the 14th to 18th days of intrauterine life, with subsequent failure in the development of the transverse septum of the diaphragm and the failure in the ventromedial migration of the paired mesodermal folds of the upper abdomen.[2] Through this gap, the heart and the abdominal viscera protrude, causing ectopia cordis and Gastroschisis or omphalocele**.**

Initial management usually addresses the lack of skin overlying the heart and abdominal viscus. Death may occur early in life, usually from infection, cardiac failure, or hypoxemia. [4] Surgical therapy for neonates without overwhelmingly severe cardiac anomalies consists of covering the heart with skin without compromising venous return or ventricular ejection. Repair or palliation of associated defects is also necessary.

This case report seeks to create awareness of this condition, emphasize the need for appropriate counselling in our environment, and also review the literature on the outcome of previously reported cases.

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Cases Presentation

Baby I.B., a 5-hour-old term female neonate, presented at the Children Emergency Center of our facility following a referral from another facility on account of an exposed heart (ectopia cordis) with multiple congenital abnormalities noticed at birth. The baby was delivered to a 41-year-old P4+1 (4A) mother via emergency cesarean section on account of antepartum hemorrhage. Mother had several episodes of antepartum hemorrhage due to a low-lying placenta, which was diagnosed at 4-month gestation and managed

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conservatively. There was no history of smoking, use of herbal concoctions or unprescribed medications. There was no family history of cardiac disease, congenital anomalies, or known genetic disorder, and exposure to irradiation was ruled out. Mother had a positive history of miscarriage in the immediate preceding pregnancy. At delivery, baby cried well at birth. Birth weight was 2,100g. She was referred on account of the multiple anomalies.

Clinical Findings

On examination at presentation, she had a lower sternal cleft, ectopia cordis, anterior diaphragmatic hernia, an epigastric omphalocele, limb abnormalities, and dysmorphic facies (flat nasal bridge and low set ears - Fig. 1). Limb abnormalities noted were an absence of the right midleg and foot (stump with a blind-ended sinus), hypoplastic left first toe, and syndactyly of the 2nd and 3rd digits of the left hand. She was tachypnoeic with a respiratory rate of 75 cycles per minute and a peripheral oxygen saturation of 91%. She was placed on intra-nasal oxygen. Other examination findings were not remarkable. She was subsequently admitted into the neonatal intensive care unit, and the eviscerated heart and omphalocele were dressed in normal saline-moistened gauze and sofra-tulle, respectively.

Diagnostic Assessment

A complete blood count and serum procalcitonin were suggestive of sepsis. She also developed hyponatremia (120 mmol/L), hypokalemia (2.5 mmol/L), and moderately elevated hyperbilirubinemia while on admission. Imaging studies, such as a thoracoabdominal computed tomography (CT) scan and echocardiography, were requested but were not done before her demise due to severe financial constraints.

Interventions Given and Outcome

The eviscerated heart was dressed three times daily with gauze moistened with normal saline, while the omphalocele was cleaned daily and dressed with sofra-tulle. She was also placed on antibiotics. Her management was multidisciplinary. Pediatric cardiologists, cardiothoracic, orthopedic, and pediatric surgeons reviewed her. The hyponatremia, hypokalemia, and neonatal jaundice were corrected with appropriate fluids and treated with phototherapy, respectively. Her parents were extensively counseled on the condition, the possible developmental failure, treatment options, and our palliative line of management which would include covering the exposed heart with skin flaps, and the likely prognosis. As the patient was unable to afford echocardiography before demise we were not able to counsel on any corrective cardiac surgery options. The baby died on the 9th day of life before the palliative surgical intervention could take place. Post-mortem reported both cardiac and extra-cardiac defects, which include ectopic heart on the anterior chest wall, absent pericardium, ventricular septal defect (0.9cm in diameter) with overriding aorta, and absence of part of the anterior diaphragm amongst other anomalies. Atelectasis and cerebral edema which were likely complications of mediastinal injuries, sepsis, and electrolyte imbalance were also reported.

Figure 1. Pentalogy of Cantrell: Clinical photographs of the patient

Discussion

POC is a very rare structural congenital syndrome presenting with five classic features, and the index patient had all five features.[5,6] The condition may be diagnosed at the first-trimester ultrasound. The prenatal diagnosis allows families to make informed decisions regarding further pregnancy management. Additionally, a definitive intervention can also be planned with this information.[7] Antenatal diagnosis was never made in our patient, though the mother had several antenatal ultrasounds. The accuracy of antenatal diagnosis for POC may depend on the level of expertise of the sonographer. After birth, a chest X-ray can reveal diaphragmatic hernia and dextrocardia. The association of ectopia cordis and omphalocele is the best diagnostic clue for this syndrome, which was present in our patient. She was a complete variant and had other structural defects; limb abnormalities, and dysmorphic facies. Other important differentials to POC are the limb body wall complex, body stalk anomaly, and the amniotic band syndrome. Unlike POC these other anomalies are more associated with the involvement of the anomalies of the face, central nervous system, and the limbs.[3]

Craniofacial defects and limb-body wall anomalies have been associated with severe and fatal variants.[7] Reported findings in the limb body wall complex include a short umbilical cord, craniofacial defects, and amniotic band anomalies. [8] Also, majority of the cases described in the literature underwent fetal demise. [8] Our patient had no obvious craniofacial abnormality and had the classical components of POC.

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Postnatal investigations in POC should include imaging studies to evaluate the extent of the respective defects to aid in planning for surgical corrective procedures. Though our patient could not access these imaging studies due to financial difficulties, a CT scan and echocardiography are necessary for the evaluation. The treatment of the POC is tailored towards the respective defects and their extent. Reconstructive repairs can be single or multi-staged and require an interprofessional team approach.[3] Surgical management of cardiac, diaphragmatic, and other defects is crucial and initially involves covering the defects.[3] Our patient remained unstable till her unfortunate demise and could not undergo the surgical interventions.

The survival rate of patients with this condition is around 37%.[9] The prognosis of POC depends on the severity of intra and extra-cardiac defects, pulmonary hypoplasia, the extent of abdominal wall defect, cerebral anomalies, and diaphragmatic herniation. The mean survival rate without any interventional surgery is about 36 hours.[10] Studies showed that even with care monitoring in professional centers and multiple corrective surgeries, they had high morbidity and mortality rates,[7] and the long-term prognosis was poor.[11] Additionally, the involvement of facial dysmorphism and limb-wall complexes confer poor prognosis.

Counselling is an important aspect of managing infants with congenital anomalies as the affected families are usually anxious and distressed. [12] Many perceive it as a calamity that had befallen them. Multi-stage counselling may be considered like in our patient. Giving too much negative information at once may tip the caregivers into losing hope or worse, abandonment of the baby in our environment. [13] However, to make progress in doing what is best for the affected child, ensuring the caregivers understand the diagnosis, and treatment options cannot be overemphasized.

Conclusions

Though rare, POC can occur in our environment. The index patient presented with the classical features in addition to limb-wall anomalies. The morbidity course and fatal outcome supported previous findings that the severity of extra and intracardiac defects leads to worse outcomes. Based on documented poor neonatal outcomes, there is a need to emphasize appropriate antenatal and postnatal assessment to provide effective counselling on termination, neonatal palliative care, and surgical repairs as appropriate.

Author Contribution

All authors contributed to the management of the patient and made significant contributions to the writing of the manuscript. BNE, FO, and OM conceived the idea of the manuscript and its design. BNE, FO, FA, IA, AOA, and SIO did the literature search, collected data, and drafted the manuscript. IBF and VCE Supervised the work. All authors reviewed the manuscript, provided critical feedback, and helped shape the final draft. All authors read and approved the final manuscript submitted

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