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Case Report

Complete pentalogy of Cantrell associated with ectopia cordis and multiple anomalies: A case report from a low-resource setting ☆

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ABSTRACT

This report presents a case of a male neonate diagnosed with a complete (Class I) PC characterized by a midline anterior wall defect with herniation of the heart and abdominal organs (ectopia cordis and omphalocele), along with diaphragmatic, sternal, and ventral abdominal wall defects consistent with PC. Additional anomalies included alobar holoprosencephaly, spina bifida, and clubfoot. The neonate was delivered via preterm cesarean section due to poor prognosis associated with this PC and other structural anomalies that were detected. The report explores the potential for associated anomalies in PC and examines the overall prognosis for individuals with PC, particularly within a low-resource setting. The case highlights managing this complex congenital condition.

Introduction

Pentalogy of Cantrell (PC) is an uncommon congenital condition that carries a high risk of serious health complications and mortality. The condition is estimated to appear in roughly 1 to 65 cases for every 200,000 live births [1]. The syndrome was first characterized and described by Dr. Cantrell in 1958, leading to its eponymous designation as "Pentalogy of Cantrell." A hernia in the diaphragm, an omphalocele (abdominal wall defect), a sternal cleft, a defect in the pericardium, and cardiac malformations are the 5 major birth defects that form the

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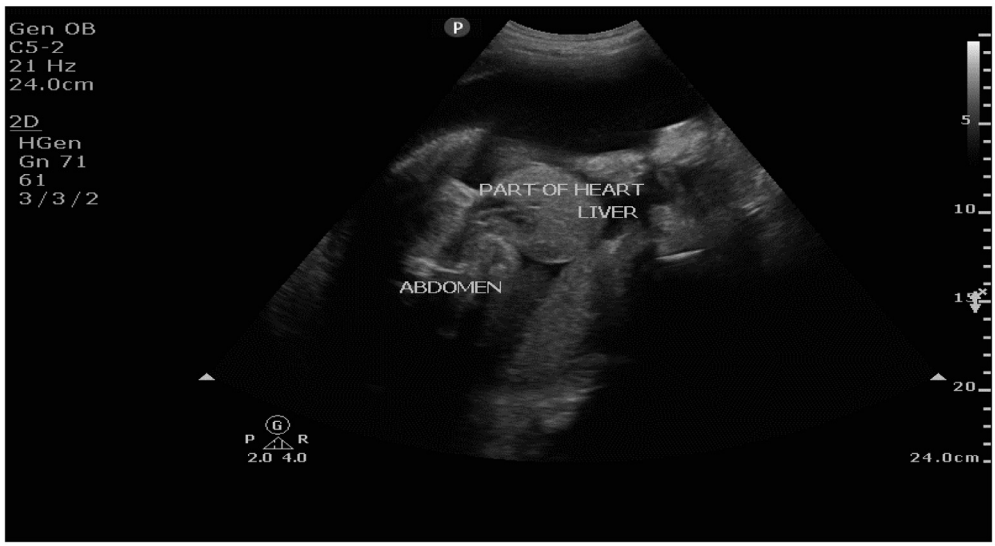

Fig. 1 – A 2-dimensional sonogram showing a membrane-covered heart and other abdominal organs including the liver extruding out of the lower sternum and upper abdomen.

condition, hence the name "pentalogy" [2]. The prevailing hypothesis regarding the etiology of PC suggests a disruption in the development of the lateral mesoderm during the embryonic period, specifically between days 14 and 18. While there is no clear genetic cause identified for the syndrome, studies are showing that it often appears alongside other genetic conditions, like Edwards syndrome and Goltz-Gorlin syndrome [3,4]. In addition, clinical reports have noted that people with PC may also exhibit other structural abnormalities, including craniofacial features like cleft palate and extra nasal openings, central nervous system issues such as hydrocephalus and neural tube defects, as well as various skeletal and additional abdominal malformations [5]. Furthermore, a small number of documented cases have reported the presence of ectopia cordis in conjunction with the full spectrum of PC [6]. Here, we present a case of PC associated with ectopia cordis and other abnormalities in a male newborn diagnosed in the third trimester and delivered by the caesarian section at our health facility.

Case report

A 19-year-old patient gravida 1, para 0 came to our department for prenatal anomaly ultrasound examination at 30 weeks + 4 days on account of polyhydramnios and suspected fetal anomalies. Apparently, that was the first scan she took upon conception. The patient had no significant medical history, no family history of congenital anomaly and was not a regular Antenatal Care attendant. She denied tobacco or alcohol use.

Ultrasound revealed a single, live intrauterine gestation corresponding to a gestational age of 30 weeks and 6 days. A membrane-covered midline anterior wall defect was identified between the lower sternum and umbilicus, containing herniated cardiac and abdominal organs, including the liver and bowels (Fig. 1). This finding, in conjunction with diaphragmatic and ventral abdominal wall defects, was consistent with omphalocele and ectopia cordis. Other fetal abnormalities, such as alobar holoprosencephaly (Fig. 2), spina bifida (Fig. 3), and clubfoot (Fig. 4) was also revealed on ultrasound. Procedures such as chorionic villus sampling, chromosomal microarray, and amniocentesis weren't performed due to limited resource availability. Financial constraints prevented the use of fetal MRI or 3D ultrasound. Fetal echocardiography was also not conducted to evaluate for intracardiac anomalies. Based on the identified ultrasound findings, a diagnosis of PC was established. Given the significant risks associated with PC and the presence of additional fetal anomalies, a multidisciplinary approach was implemented for the patient's management. A team of obstetricians, gynecologists, and counsellors collaborated to develop a comprehensive care plan. Following careful conversations with the patient and her family, a decision was reached to move forward with a Cesarean section. This choice was made due to the fetus's poor prognosis and the potential risks to the mother if the pregnancy continued. Throughout this period, the patient received both counseling and emotional support.

Intraoperative findings at 31 weeks of gestation revealed a live male infant in a cephalic presentation, with several congenital abnormalities, such as the absence of the neck and shoulders, omphalocele with ectopia cordis, and spina bifida (Fig. 5). The neonate weighed 1.0 kg at birth and had Apgar scores of 4 at 1 minute and 7 at 5 minutes. The head circumference was measured at 27 cm.

Discussion

PC is an uncommon and intricate congenital condition marked by 5 main abnormalities: a diaphragmatic hernia, an abdominal wall defect (omphalocele), a sternal cleft, a defect

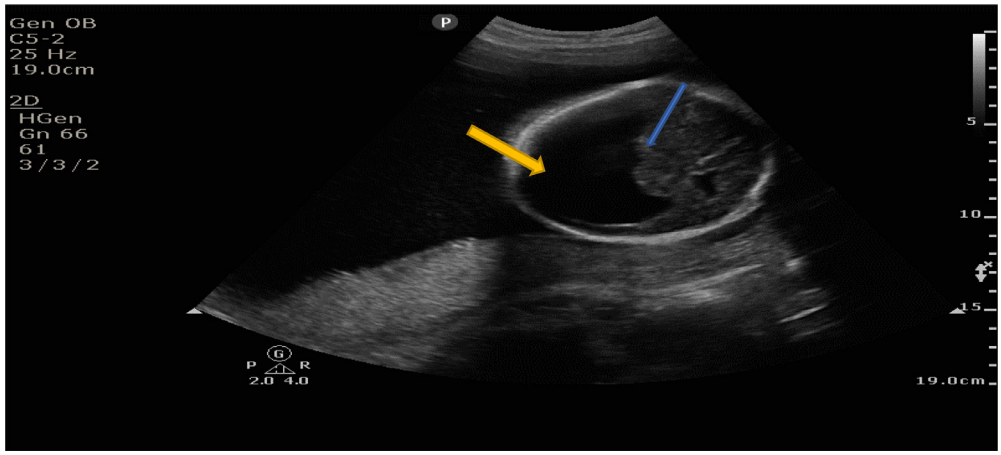

Fig. 2 – A 2-dimensional sonogram of the fetal brain showing alobar holoprosencephaly with fused thalami (blue arrow) and dilated mono-ventricle (yellow arrow).

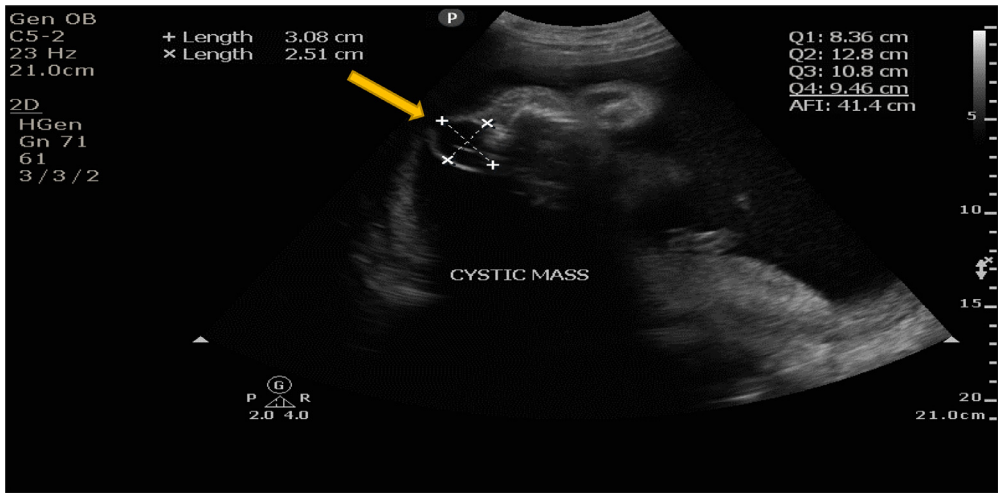

Fig. 3 – Spina bafida: A 2-dimensional sonogram of the fetal dorsal region shows a complex cystic lesion at the cervico-thoracic region.

in the pericardium, and various heart malformations. This report discusses the case of a 19-year-old woman in her first pregnancy, who, at 30 weeks gestation, was found to be carrying a fetus with a complete form of PC, along with other anomalies such as alobar holoprosencephaly, spina bifida, and clubfoot. The diagnosis was established through prenatal ultrasound examination, which revealed significant structural abnormalities, including ectopia cordis and omphalocele, consistent with the diagnostic criteria for PC. PC is an uncommon condition, with studies indicating prevalence rates ranging between 1 in 65,000 and 1 in 200,000 live births [7,8]. The present case aligns with the established understanding of PC as a primarily sporadic occurrence, with no family history or known genetic predisposition identified. While most cases are considered sporadic, previous literature has documented instances of PC associated with chromosomal abnormalities, including Trisomy 18, Trisomy 21, and Turner Syndrome [9]. Chromosomal testing, including chorionic villus sampling and amniocentesis, was not performed due to the facility's limited resources. This precluded the possibility of identifying any potential underlying genetic factors that may have contributed to the development of the PC and associated anomalies in this specific case.

The presence of ectopia cordis, in this case, aligns with established research, which has consistently identified ectopia cordis as a hallmark feature of severe PC. Cakiroglu et al.,[10] documented a similar case presenting with ectopia cordis alongside other PC-related defects, resulting in unfavorable neonatal outcomes. Consistent with Cakiroglu's findings, this case also exhibited a poor prognosis, as evidenced by low Apgar scores at birth (4 at 1 minute and 7 at 5 minutes) and severe anomalies incompatible with long-term survival.

The clinical presentation of this case is notable for the association of PC with additional anomalies, including alobar holoprosencephaly, spina bifida, and clubfoot, which are not commonly reported in conjunction with this syndrome. Previous studies such as, Grigore et al.,[11] have acknowledged the possibility of additional craniofacial and skeletal

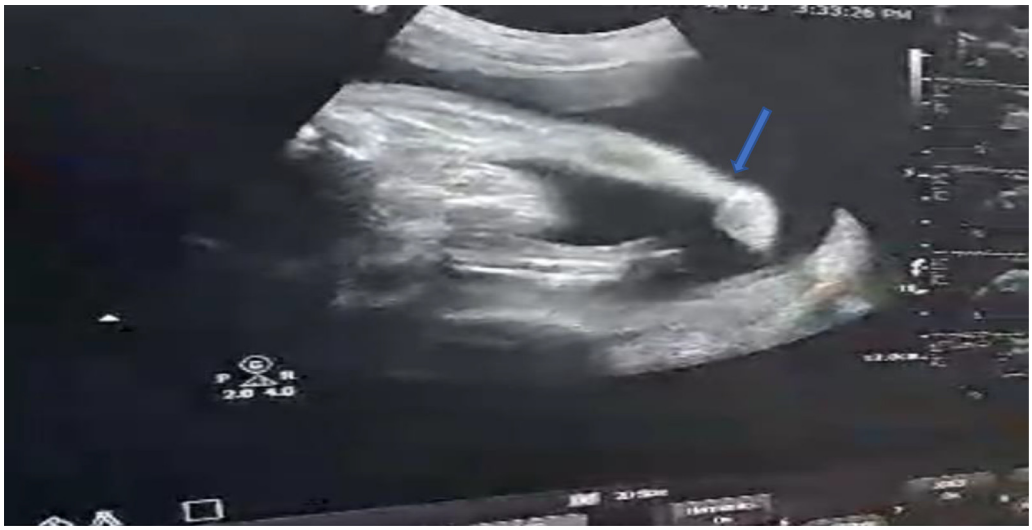

Fig. 4 – Clubfoot: A 2-dimensional sonogram of the fetal lower extremity showing inversion and medial rotation of the foot (blue arrow).

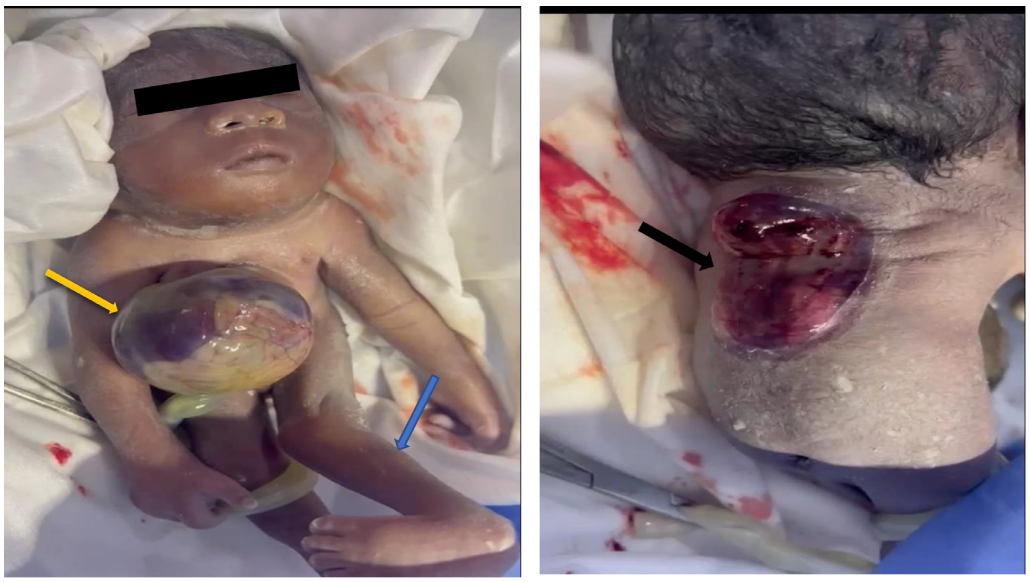

Fig. 5 – Postdelivery images show a neonate with extrusion of the heart and abdominal organs including the liver and bowels (yellow arrow), club foot (blue arrow) and Spina Bifida (black arrow).

anomalies in PC cases, although these are not consistently present. The presence of alobar holoprosencephaly, a severe brain malformation, in this case significantly complicated the prognosis and management, suggesting a particularly severe disruption in early embryonic development.

Previous research has classified PC into complete and incomplete forms, based on the presence of all 5 defining defects or a subset of them, respectively. Toyama's classification system for PC categorizes cases into 3 distinct groups. The present case aligns with the "definite" (Class 1) category, as it exhibits all 5 characteristic defects of PC, in addition to presenting with additional structural anomalies [12]. The presence of alobar holoprosencephaly and other neural tube defects in this case suggests a more widespread disruption in embryonic development than typically described in the original classification of PC. This case contributes to the growing body of evidence that the PC can co-occur with severe central nervous system abnormalities, further complicating the clinical presentation and significantly reducing the likelihood of survival.

The outcome of this case aligns with previously reported data, which consistently demonstrates a significantly low survival rate for infants with PC, particularly when ectopia cordis is present [13]. Surgical correction of defects associated with

PC poses significant challenges. It is often only feasible in cases with less severe cardiac malformations and the absence of ectopia cordis. Harring et al.,[14] highlight the exceptionally low survival rate in cases with complex intracardiac defects, such as those observed in the current case. Financial constraints and limited access to advanced prenatal diagnostic tools, such as fetal echocardiography and MRI, hindered the full evaluation of the intracardiac defects in this case.

PC, a multifaceted congenital disorder, presents significant diagnostic and management complexities. Although prenatal ultrasound remains the primary tool for diagnosis, advanced imaging methods like fetal MRI, 3D/4D ultrasound, and fetal echocardiography provide essential details on the severity of the condition and any related anomalies [15,16]. Effective management mandates a multidisciplinary approach, encompassing prenatal counseling, meticulous delivery planning, and staged surgical interventions to address cardiac, abdominal, and sternal defects [17,18]. Cesarean delivery at a tertiary care center is often recommended, followed by timely surgical repair of life-threatening defects. Long-term care is paramount for survivors, given the potential for developmental delays and other complications. While severe cases often have a poor prognosis, early detection and focused care can enhance the potential for improved outcomes. With proactive management, we can foster a more positive path forward.

Conclusion

This case study highlights the poor prognosis of PC, especially in severe cases with ectopia cordis and additional anomalies such as neural tube and cranial defects, which complicate management. Early diagnosis and multidisciplinary care are crucial, yet, as reported in prior research, neonates with complete PC face unfavorable outcomes. This case is particularly insightful for low-resource settings, showcasing the full severity of PC alongside extra anomalies, and emphasizing its complex, multifactorial origins. While multidisciplinary care was applied, the neonatal survival outcome aligns with the historically poor prognosis for complete PC cases.

Patient consent

Written informed consent was obtained from the patient prior to the publication of this case report.

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