PRENATAL DIAGNOSIS OF PENTALOGY OF CANTRELL WITH ABSENT LIMB: A RARE CASE REPORT FROM ETHIOPIA

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ABSTRACT

BACKGROUND: Pentalogy of Cantrell is rare and serious condition that affects roughly 5.5 out of every 1,000,000 live newborns. It comprises five characteristics defects: omphalocele, cardiac ectopia, absences of the anterior diaphragm, the lower sternum, and of the parietal diaphragmatic pericardium. We report a case of severe form of pentalogy of Cantrell categorized as class 1 having all five of the diagnostic defects and associated with absent upper left limb (amelia).

CASE PRESENTATION: A 21-year-old primigravida mother with 8 months amenorrhea presented at our hospital for routine antenatal care visit. A detailed obstetric ultrasound scanning was performed and revealed characteristic findings of pentalogy of Cantrell A multidisciplinary team discussed with the mother and the family about the severity of the fetal anomaly as well as its poor prognosis and the pregnancy was terminated medically. A 1600gm male newborn was delivered vaginally which that later passed away after 10 minutes with no maternal complication.

CONCLUSION: In the context of complicated pentalogy of Cantrell, this case highlights the vital importance of prompt prenatal screening, thorough counselling, and individualized management planning by multidisciplinary team.

KEYWORDS: Pentalogy of Cantrell, Ectopia cordis, Omphalocele, Amelia, Case report, Ethiopia

(The Ethiopian Journal of Reproductive Health; 2026; 18; 79-84)

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INTRODUCTION

The first description of the Cantrell pentalogy was published in 1958. It is severe and rare congenital condition characterized by five anomalies: a midline supraumbilical abdominal wall defect; lower sternal defect; anterior diaphragmatic defect; diaphragmatic pericardial defect, and congenital abnormalities of the heart[1]. Its incidence is about 5.5 out of every 1,000,000 live newborns and more prevalent in male gender than female which is nearly three times higher[2]. Globally, the prevalence of congenital anomaly is about 3-6% of births, while in Africa varies between countries and its pooled estimates are between 2-3%[3,4].

Here we reported a class 1 category of pentalogy of cantrell (characterized by a midline defect, upper abdominal wall abnormality; lower sternal defect; anterior diaphragmatic defect; and diaphragmatic pericardial defect) associated with absent limb which is rarely reported case globally and challenging to diagnose in resource- limited settings like Ethiopia.

Case Presentation

A 21 –year- old primigravida presented after eight months of amenorrhea, referred from a nearby General Hospital with the diagnosis of omphalocele and 3rd trimester pregnancy in her third antenatal visit for further evaluation and management. The mother had two ANC contacts starting from four months of amenorrhea. She has no family history of fetal congenital anomalies and took folate and iron supplementation during the current pregnancy. The mother is housewife farmer and had no history of any use of medication. Physical examination revealed, normal vital signs, pink conjunctiva, a 30-week-sized gravid uterus, longitudinal lie, cephalic presentation, and positive fetal heartbeat. Other physical exam findings were unremarkable.

Diagnostic Assessment

Obstetric ultrasound scan done by Feto-Maternal Medicine (FMM) confirmed singleton intrauterine pregnancy at 31week plus 4 days, positive fetal heartbeat, fundal - posterior placenta, large abdominal wall defect with floating of liver, bowel loops and kidney in the amniotic fluid with no covering membrane, heart is located completely outside the thoracic cavity floating in the amniotic fluid, and single deepest pocket is 8.5cm. This conclusion from the ultrasound features was: 3rd trimester pregnancy + pentalogy of Cantrell +mild polyhydramnios (see Figure 1).

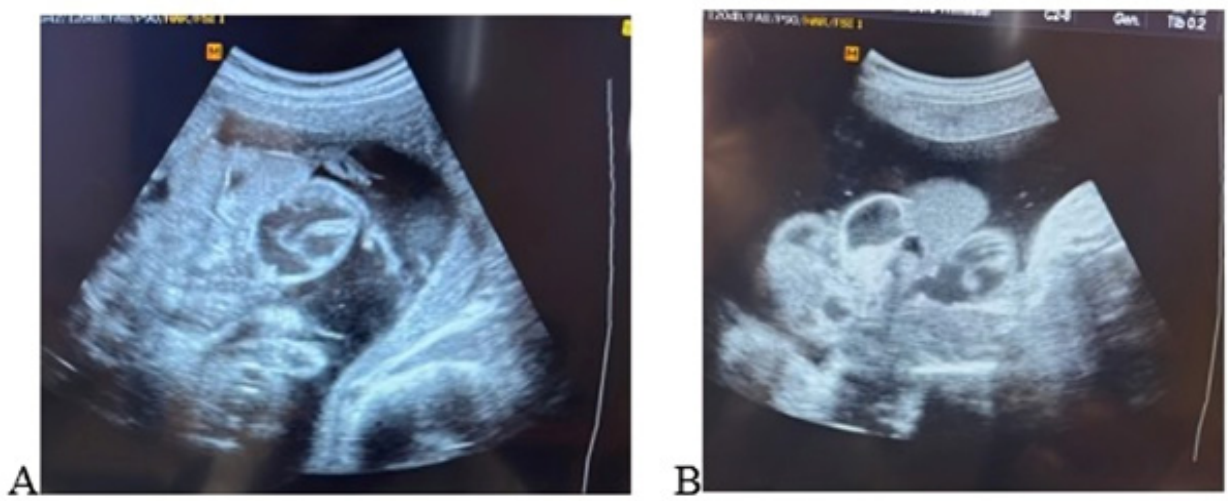

Figure 1: Tran-abdominal obstetric ultrasound A (heart floating in the amniotic fluid), B (liver and bowel outside abdominal cavity)

Investigations were done and all were in the normal range (see Table 1).

Table 1: Details of laboratory investigations

Lists of investigations	Dates		Remark
	June 24, 2025	June 30, 2025	
Random blood sugar (mg/dl)	95		
Complete blood count			
White blood cell (count/Ul)	13.1*103	17.2*103	
Hemoglobin (g/dl)	12.4	12.7	
Hematocrit (%)	35.9	38.3	
Platelet (count/uL)	290*103/	283*103	
VDRL	Negative		
HBSAg	Negative		
Urinalysis	Nonrevealing		
Blood group	O		
Rh status	Negative		
Indirect coombs test	Negative		

NB: mg/dl: milligram per decilitre, g/dl: gram per decilitre, %: percent, Ul: unilitre, VDRL: Venereal Disease Research Laboratory, HBSAg: Hepatitis B surface antigen, Rh: Rhesus

Therapeutic Intervention and Outcomes

After the diagnosis was confirmed the prognosis of the pregnancy was discussed with the mother and family thoroughly about the lethal nature of the anomaly and its prognosis. She was admitted to maternity ward for pregnancy termination. Cervical ripening was done with 25micg misoprostol three doses and induction of labor with Pitocin following the protocol of hospital OBGYN department. A 1600gm male newborn was delivered vaginally that later passed away after 10 minutes with no maternal complication.

Then the neonate was evaluated with senior physician and the findings were as follows: there is midline anterior abdominal wall defect extending from umbilicus to lower third of sternum through which liver, bowel lops and heart are eviscerated with no covering membrane. The newborn is born with absent left upper extremity (see Figure 2 below).

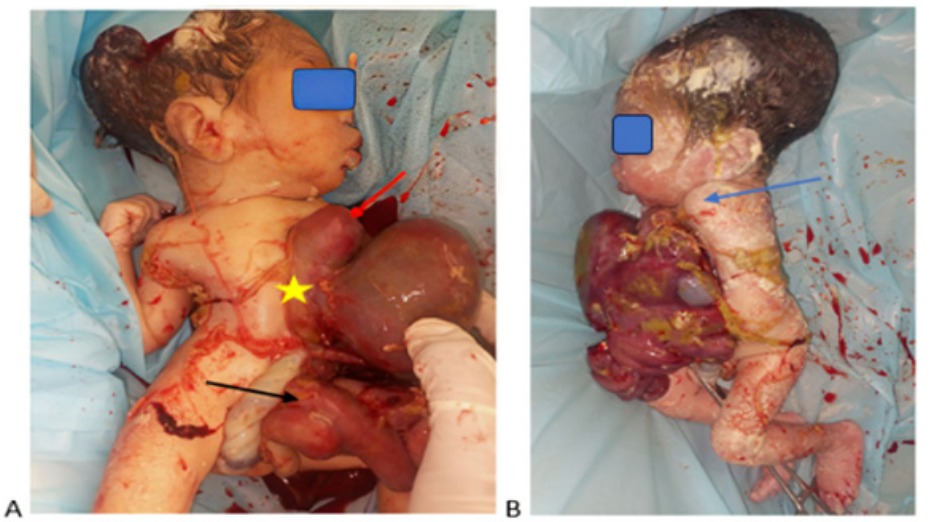

Figure 2: pictures of the neonate immediately taken after delivery: A, eviscerated thoraco-abdominal organs: bowel loops (black arrow), liver (yellow star), heart (red arrow) and B, Absent left upper extremity (blue arrow).

DISCUSSION

In 1958, Cantrell was the first to identify the pentalogy of Cantrell, a severe and rare congenital condition characterized by five anomalies: a midline, upper abdominal wall abnormality; lower sternal defect; anterior diaphragmatic defect; diaphragmatic pericardial defect, and congenital abnormalities of the heart[1]. Its incidence is about 5.5 out of every 1,000,000 live newborns live births and the prevalence is nearly three times higher in men than in women[2,5].

Cantrell pentalogy's precise etiology is unknown; however, it is most likely complex, involving a mix of environmental and genetic variables[6]. It is hypothesized that a section of the lateral mesoderm failed to form during 14–18 days of gestation and that the rupture of the yolk sac and chorion prevented the chest wall from properly fusing in the midline[1,7]. Genetic factors implicated in the development of pentalogy of Cantrell include mutations in BMP2 (bone morphogenetic protein2), which is responsible for the normal development of midline structures, and ALDH1A2, which is crucial for the conversion of vitamin A to retinoic acid, which plays a major role in organogenesis, pleuroperitoneal folding, and diaphragmatic embryogenesis[1,8,9]. The majority of Cantrell pentalogy cases are sporadic. There have been few documented cases linked to trisomy 18 and X linked inheritance [10,11].

In 1972, Toyama divided the Cantrell pentalogy into three categories 12. Class 1 (definitive diagnosis), which exhibited all five significant defects and additional primary and minor anomalies. Class 2, probable diagnosis, with four defects present, including intracardiac and ventral wall abnormalities; and class 3, incomplete expression, with various combinations of defects present, including a sternal abnormality. Our case report has all five defects which definitely categorized as class 1.

Malformations can occasionally be so subtle that they are challenging to detect, even after delivery. In 80% of cases, ectopia cordis is frequently linked to this syndrome[6,13,14]. For the screening and diagnosis of pentalogy of Cantrell, ultrasound is a vital tool. Pentalogy of Cantrell has been diagnosed by sonography as early as 10–12 weeks of pregnancy, and it should be suspected if an omphalocele and an ectopia cordis are found 15. While high-resolution 2D ultrasound is just as effective as 3D in the early stages of pregnancy [5], employing 3D ultrasound as an adjuvant may improve the visibility of fetal abnormalities in several orthogonal planes 16. In our case we detected all five anomalies on prenatal ultrasound despite the late presentation in the 3rd trimester pregnancy which makes it different from previous case reports from Ethiopia that were diagnosed postpartum[17,18].

Critical anatomic features, measurements, and structural information all benefit from three-dimensional CT reconstruction. MRI provides the best evaluation of this syndrome by characterizing ultrasound results and providing additional diagnostic details[19]. Additionally, when there is significant oligohydramnios, which makes it impossible to see the fetal parts clearly with ultrasound, MRI can be very beneficial[20]. But in our case, we didn't offer CT scan and MRI imaging modalities since ultrasound features were typical of pentalogy of Cantrell. The diagnosis is confirmed during postnatal examination (after delivery).

Numerous related abnormalities, such as limb malformations like tibia and radius absence, hypodactyly, and even phocomelia, as well as craniofacial and central nervous system abnormalities such cleft lip and palate, encephalocele, hydrocephalus, and craniorachisis have been documented in the literature[21,22]. In our case it is associated with phocomelia (absent left upper arm).

Pentalogy of Cantrell is mostly managed based on the severity of the malformations and related abnormalities. The majority of cases carry an extremely poor prognosis, and it is recommended that the pregnancy be terminated. Because of the thoracic cage's hypoplasia, which makes it impossible to contain the ectopic heart,

surgically correcting the abdominal wall defect is frequently challenging in complex situations[9,23]. A multidisciplinary team must establish the optimal timing of birth and appropriate surgical therapies for mild forms of pentalogy Cantrell. Clearly, those with mild abnormalities have better results[11,24]. In our case since the prognosis is poor the termination of pregnancy was determined after a thorough discussion with the family.

Conclusion

In the context of complicated pentalogy of Cantrell, this case highlights the vital importance of prompt prenatal screening, thorough counselling, and individualized management planning. We recommend the hospitals to make anatomic scan at 18 to 22 weeks as a routine service pregnant mother.

Ethical Approval

We conducted the case report in compliance with the Declaration of Helsinki, Good Clinical Practices, institutional regulatory requirements. Jimma University Medical Centre Ethical Committee approved it.

Consent for Publication

The patient signed informed consent forms for the use of case details and images for publication and for scientific purposes.

Acknowledgments

The authors thanked to all multidisciplinary team involved in the management of the case and the mother and her family for their consent to publish the case.

Author Contributions

All authors made a significant contribution to the work reported, whether that is in the conception, study design, execution, acquisition of data, analysis and interpretation, or in all these areas; took part in drafting, revising or critically reviewing the article; gave final approval of the version to be published; have agreed on the journal to which the article has been submitted; and agree to be accountable for all aspects of the work

Disclosure

The author reports no conflicts of interest in this work.

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REFERENCES

1. Cantrell JR, Haller JA, MM R. A syndrome of congenital defects involving the abdominal wall, sternum, diaphragm, pericardium, and heart. Surgery, Gynecology & Obstetric. 1958;107(5):13.
2. Jnah AJA, DNP, NNP-BC; Newberry, Desi M. APRN, DNP, NNP-BC; England, Amanda MD. Pentalogy of Cantrell: case report with review of the literature. . Advances in Neonatal Care. 2015;15(4):8.
3. Moges N, Anley DT, Zemene MA, et al. Congenital anomalies and risk factors in Africa: a systematic review and meta-analysis. BMJ Paediatrics Open. 2023;7(1):e002022.
4. Jemal S, Fentahun E, Oumer M, Muche A. Predictors of congenital anomalies among newborns in Arsi zone public hospitals, Southeast Ethiopia: a case-control study. Italian Journal of Pediatrics. 2021;47(1):143.
5. Zvizdic Z S-PI, Mesic A, Terzic S, Vranic S. The complete spectrum of pentalogy of Cantrell in one of a set of dizygotic twins: a case report of a rare congenital anomaly. Medicine. 2012;100(14):25470.
6. Fazea M, Alhameli M, Ahmed F, et al. Pentalogy of Cantrell associated with ectopia cordis: a case report. Pediatric health, medicine and therapeutics. 2022:283-287.
7. Williams AP MR, Beierle EA. . Pentalogy of cantrell. In Seminars in pediatric surgery. WB Saunders. 2019;28(2):16.
8. Adane L, Bedane A, Abera A. MRI Findings in Pentalogy of Cantrell with Ultrasound Correlation: Addis Ababa, Ethiopia. Gynecol Reprod Health. 2018;2(2):1-5.
9. Mayilvaganan KR, Prathyusha IS, Gautam MS, Raidu D, VN A. A recurrent case of Pentalogy of Cantrell: a rare case with sonological findings and review of literature. Polish Journal of Radiology. 2017;82:28-31.
10. Yamasaki A, Pedreira D, Muller R, Machado M, Schultz R, Cha S. P115First trimester diagnosis of the Pentalogy of Cantrell. Ultrasound in Obstetrics and Gynecology. 2000;16:91-91.
11. Williams AP, Marayati R, Beierle EA. Pentalogy of cantrell. Paper presented at: Seminars in pediatric surgery2019.
12. 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):778-792.
13. de Rubens Figueroa J, Cruz EFS, García LD, Daza DC. Malformaciones cardiacas en pacientes con pentalogía de Cantrell y ectopia cordis. Revista Española de Cardiología. 2011;64(7):615-618.
14. Madi JM, Festugatto JR, Rizzon M, Agostini AP, de Araújo BF, Garcia RMR. Ectopia Cordis Associated with Pentalogy of Cantrell—A Case Report. Revista Brasileira de Ginecologia e Obstetrícia/RBGO Gynecology and Obstetrics. 2019;41(05):352-356.
15. Key R. Pentalogy of Cantrell. 2019. Accessed July 6, 2025.
16. Rodgers EB, Monteagudo A, Santos R, Greco A, Timor-Tritsch IE. Diagnosis of Pentalogy of Cantrell Using 2‐and 3‐Dimensional Sonography. Journal of ultrasound in medicine. 2010;29(12):1825-1828.
17. Tadele H, Chanie A. Thoracic ectopia cordis in an Ethiopian neonate. Ethiopian journal of health sciences. 2017;27(2):203-205.
18. Ayalew S, Negussie MA, Afework HT, Kassie DA, Mogess AM, Wassie MT. Pentalogy of Cantrell in a resource-limited setting: A case report. Journal of Pediatric Surgery Case Reports. 2025;112:102935.
19. Leyder M, Van Berkel K, Done E, Cannie M, Van Hecke W, Vorsselmans A. Ultrasound meets magnetic resonance imaging in the diagnosis of pentalogy of cantrell with complete ectopy of the heart. Gynecol Obstet (Sunnyvale). 2014;4(200):2161-0932.1000200.
20. Said AH, El-Kattan E, Abdel-Hakeem AK, Saleem S. In utero MRI diagnosis of fetal malformations in oligohydramnios pregnancies. The Egyptian Journal of Radiology and Nuclear Medicine. 2016;47(4):1733-1742.
21. Jagtap SV, Shukla DB, Jain A, Jagtap SS. Complete Pentalogy of Cantrell (POC) with Phocomelia and Other Associated Rare Anomalies. Journal of clinical and diagnostic research : JCDR. May 2014;8(5):Fd04-05.
22. Morales JM, Patel SG, Duff JA, Villareal RL, Simpson JW. Ectopia cordis and other midline defects. The Annals of thoracic surgery. 2000;70(1):111-114.
23. Oguk F SL, Ongeti C, Ngeno S, Kipchirchir C, Ndiangui F, Nalianya W, Sitters J, Onditi E, Rahmani R (2015); ANTENATAL DIAGNOSIS OF PENTALOGY OF CANTRELL IN EAST AFRICA: A CASE REPORT Int. J. of Adv. Res. 3 (Mar). 0] (ISSN 2320-5407.
24. Júnior EA, Carrilho MC, Toneto BR, Guilhen JCS. Pentalogy of Cantrell: prenatal diagnosis, delivery, and immediate postnatal surgical repair. Journal of neonatal surgery. 2017;6(2):32.