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Complete Thoracic Ectopia Cordis without an Associated Abdominal Wall Defect: A Case Report of Diagnostic and Therapeutic Challenges in a Sub-Saharan African University Hospital

Waaviel Carine Linda Somé[1], Zénabou Boussini[1*], Alihonou Serges Eric Togbe[2,3], Fatimata Zongo[4], Semon Paulin Kam[1], Jean De la Croix Millogo[1,3], Evelyne Komboigo[3,5], Adama Dembélé[1,3], Der Adolphe Somé[1,3]

[1]Department of Obstetrics and Gynaecology, Souro Sanou University Teaching Hospital, Bobo-Dioulasso, Burkina Faso
[2]Department Obstetrics and Gynaecology, Medical Center with Surgical Antenna of Do, Bobo-Dioulasso, Burkina Faso
[3]Faculty of Health Sciences, Nazi Boni University, Bobo-Dioulasso, Burkina Faso
[4]Department of Pediatrics and Neonatology, Souro Sanou University Teaching Hospital, Bobo-Dioulasso, Burkina Faso
[5]Department of Obstetrics and Gynaecology, Pala University Teaching Hospital, Bobo-Dioulasso, Burkina Faso
Email: *zenaboussini@yahoo.fr

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Abstract

Background: Ectopia cordis is a rare congenital malformation with a very poor prognosis that varies with associated anomalies and access to specialized treatment. Late prenatal diagnosis and lack of specialized pediatric cardiothoracic surgery at the treating institution exacerbate clinical and ethical challenges. **Case:** A 29-year-old multiparous woman was referred at 37 weeks gestation following ultrasound revealing complete thoracic ectopia cordis without abdominal wall defects. Planned caesarean section at 38 weeks delivered a live neonate weighing 2900 g with Apgar scores 7/8. Palliative care was provided; death occurred at 48 hours of age. **Conclusion:** This case illustrates critical diagnostic and therapeutic inequities in sub-Saharan Africa: 1) Late antenatal diagnosis, limiting opportunities for timely parental counselling; 2) Absence of pediatric cardiothoracic surgery capacity at the treating institution. We advocate for urgent expansion of obstetric imaging screening in low-resource settings.

Keywords

Ectopia Cordis, Rare Congenital Malformation, Prenatal Diagnosis, Sub-Saharan Africa, Diagnostic Delay, Therapeutic Inequity

1. Introduction

Ectopia cordis is a rare congenital cardiac malformation with an incidence of 5.5 - 7.9 per million live births [1]. It represents failure of midline fusion of mesodermal folds between weeks 3 and 4 of gestation. The externalized heart lacks pericardial and sternal enclosure, creating cardiac vulnerability.

The pentalogy of Cantrell, defined by Cantrell *et al.* in 1958 [2], involves abdominal wall defects, distal sternal cleft, anterior diaphragm defect, apical pericardium defect with pericardio-peritoneal communication, and intracardiac anomalies. Thoracic ectopia cordis represents approximately 60% of cases. When abdominal wall and diaphragmatic defects are absent on imaging, thoracic ectopia cordis is diagnosed; however, the presence of additional intracardiac or diaphragmatic structural defects cannot be confirmed or excluded without formal echocardiography and, ideally, postmortem examination [3].

Neonatal prognosis remains grave. Most affected infants die within hours to days of birth from acute cardiac decompensation or refractory hypoxemia [4]. Survival, including long-term survival, has been reported after surgery in specialized centers; prognosis varies with associated anomalies and therapeutic possibilities [5].

Diagnostic disparities characterize ectopia cordis across high- and low-resource settings [3] [4]. In high-income countries, diagnosis typically occurs in the first trimester (11 - 13 weeks) by transvaginal ultrasound, allowing informed parental discussion [1] [3]. In sub-Saharan Africa, prenatal diagnosis is often delayed or entirely absent. Systemic barriers include limited access to first-trimester imaging, insufficient operator training in fetal morphology, and underfunded screening programmes [3] [4].

We report a case of thoracic ectopia cordis diagnosed late (37 weeks) at Centre Hospitalier Universitaire Souro Sanou, Bobo-Dioulasso, Burkina Faso [6], to illustrate systemic barriers to prenatal diagnosis and specialized neonatal care in low-resource sub-Saharan Africa.

2. Case Presentation

Clinical setting and maternal history

Centre Hospitalier Universitaire Souro Sanou is a tertiary referral center in Bobo-Dioulasso, Burkina Faso, serving a catchment population of approximately 2 million across the Hauts-Bassins region. The patient was a 29-year-old multiparous woman (gravida 3, parity 2) from Bobo-Dioulasso, Burkina Faso. Medical history was unremarkable with no exposure to known teratogens. Previous obstetric deliveries (2010, 2015) were uncomplicated term vaginal deliveries. No family history of congenital malformations or genetic disorders.

Pregnancy course and prenatal diagnosis

Antenatal care in resource-limited settings typically occurs only when complications arise; this patient received no routine prenatal screening until 37 weeks.

This pattern reflects limited access to screening programmes characteristic of many sub-Saharan African maternities. At 37 weeks plus 5 days, the patient presented for obstetric ultrasound. Complete thoracic ectopia cordis was diagnosed prenatally by obstetric ultrasound (Sonosite 180, 3.5 MHz), which revealed major anterior thoracic wall defect with complete externalization of the fetal heart, regular myocardial contractions (heart rate 140 - 150 bpm), cardiac chamber size appropriate for 37 weeks, normal amniotic fluid volume, and absence of abdominal wall defects or documented diaphragmatic discontinuity (see **Figure 1** and **Figure 2**).



Figure 1. Ultrasound at 37 weeks of gestation. Sagittal view of fetal trunk in two-dimensional mode: major midline anterior thoracic wall defect, heart completely located outside thoracic cavity.



Figure 2. doppler echography image. Complete thoracic ectopia cordis: heart externalized through large midsternal defect. view in color Doppler: pulsatile flow within externalized heart.

Critical limitations of the prenatal ultrasound evaluation must be emphasized: fetal echocardiography was not performed; therefore, the presence or absence of intracardiac structural anomalies (ventricular septal defects, atrial septal defects, anomalies of the great vessels) could neither be confirmed nor excluded. Similarly, the presence of pulmonary hypoplasia could not be determined by prenatal ultrasound alone. A multidisciplinary discussion (obstetrics, neonatology, pediatrics) was conducted. Given the absence of local cardiothoracic surgical capacity and specialized neonatal support infrastructure, planned caesarean section was offered to optimize neonatal transition and allow postnatal assessment. Written informed parental consent was obtained following detailed counselling regarding the very poor prognosis in this clinical setting, the limitations of anatomical assessment, and the absence of local surgical treatment options.

Delivery and postnatal examination

Planned caesarean section at 38 weeks delivered a live female neonate weighing 2900 g, vigorous and reactive. Apgar scores: 7 at 1 minute, 8 at 5 minutes. Physical examination confirmed externalized heart through a 5 × 6 cm midsternal defect, absent parietal pericardium, intact abdominal wall, and no gross diaphragmatic defect visible on examination. The neonate was notably vigorous despite the severity of the malformation, with visible, palpable myocardial contractions. Remainder of physical examination was unremarkable, excluding syndromic features associated with Pentalogy of Cantrell (see Figure 3).



Figure 3. Birth photograph: neonatal presentation. Complete thoracic ectopia cordis at birth: externalized heart with visible pulsatile contractions, major midsternal defect, absence of pericardial and cutaneous coverage.

3. Results

Neonatal management and palliative care

The neonate was admitted to intensive care. No surgical intervention was attempted, given the grave prognosis and absence of surgical repair capacity at the

treating institution. Supportive and palliative care was provided, including:

- Thermoregulation in incubator (core temperature 37.5˚C);
- Intravenous nutrition (10% dextrose);
- Myocardial protection with sterile saline-moistened gauze dressings changed every 2 hours to prevent desiccation and infection;
- Continuous cardiac monitoring per standard neonatal resuscitation protocols [7].

Clinical course and vital signs

The exposed myocardium was visibly swollen and inflamed on initial examination. Progressive cardiorespiratory decompensation occurred despite maximal supportive care. The following vital signs and clinical observations were documented in the patient's hospital record at CHU Souro Sanou and represent the actual clinical course from birth to death (see **Figure 3**) (see **Table 1**):

Table 1. Neonatal vital signs and clinical progression from birth to 48 hours of life.

Time (h)	FC (bpm)	FR (c/min)	SpO_2 (%)	TA (mmHg)	Support	Clinical Signs
0	152	68 (irr)	72	NM	Room air	Severe cyanosis, respiratory distress
6	158	68	84	54/32	Nasal O_2	Persistent cyanosis
12	166	74	78	48/26	Mask O_2	Intercostal retractions, tachypnea
24	172	78	74	42/23	Mask O_2	Signs of congestive heart failure
36	178	82	68	36/20	Mask O_2	Terminal state, severe hypotension
48	Declining	Apnea	Falling	Unmeasurable	Assist	Asystolie, death declared

Outcome

The vital signs progression (rising heart rate and respiratory rate, falling oxygen saturation and blood pressure) reflects progressive myocardial dysfunction consistent with cardiovascular failure. At 48 hours and 15 minutes of life, the neonate developed bradycardia progressing to asystolie. Standard neonatal resuscitation was performed according to established protocol. Despite appropriate measures, no return of spontaneous circulation was achieved. Death was declared at 48 hours and 15 minutes of life.

Probable cause of death: Severe myocardial dysfunction secondary to direct exposure of cardiac myocardium, loss of pericardial containment, and myocardial inflammation and edema. The exposed myocardium was unable to maintain adequate systemic perfusion despite supportive care. Persistent hypoxemia refractory to oxygen support suggests possible underlying pulmonary pathology or right-to-left shunting through persistent fetal circulatory pathways. However, formal echo-

cardiography was not performed due to critical clinical instability; therefore, intracardiac structural anomalies that might have contributed to death could not be definitively characterized. Postmortem examination was not performed due to parental and religious considerations. Consequently, the precise anatomical substrate for death remains incompletely defined.

4. Discussion

Clinical presentation and pathophysiology

This neonate survived 48 hours, considerably longer than some reported cases [4]. The initial Apgar scores (7/8) suggest initial cardiopulmonary adaptation [5]. However, myocardial exposure rapidly induced inflammation, progressive myocardial dysfunction, and inability to maintain systemic perfusion [5]. The documented clinical progression reflects progressive myocardial failure [1] [5]. Intracardiac anatomy and diaphragmatic integrity were not fully characterized by imaging; therefore, the contribution of possible intracardiac structural defects to cardiovascular decompensation cannot be excluded [3].

Diagnostic delay and health inequity

In this case, ectopia cordis was diagnosed at 37 weeks. Earlier detection is possible when prenatal ultrasound screening and appropriate expertise are available, but the timing of diagnosis varies between patients and healthcare settings [3] [5]. This single observation cannot establish a systematic difference between high-income countries and sub-Saharan Africa. Published reports nevertheless describe barriers to prenatal ultrasound access in some resource-limited settings [4]. The following considerations highlight potential challenges rather than patterns that can be assumed to apply throughout the region:

1) Access to screening: A published case report from Nigeria illustrates difficulties in accessing prenatal ultrasound in a resource-limited setting [4]. Such observations provide context for the late diagnosis in our case, but do not establish screening coverage at CHU Souro Sanou or across sub-Saharan Africa.

2) Perceptions of prenatal screening: Individual and community perceptions may influence the use of prenatal services. However, this case does not establish the patient's reasons for delayed screening or prevailing attitudes in the wider population. The role of such perceptions would require investigation through local studies.

Ethical implications of late diagnosis and Burkina Faso legal framework

Late diagnosis exacerbates ethical challenges: inability to afford genuine parental autonomy and unnecessary neonatal suffering [8]. Burkina Faso's Penal Code (Article 513-14) addresses this issue [9]. The statutory language provides (in French): "L'interruption volontaire de la grossesse peut à tout âge gestationnel être pratiquée si un médecin atteste que le maintien de la grossesse met en péril la santé de la femme ou qu'il existe une forte probabilité que l'enfant à naître soit atteint d'une maladie ou d'une infirmité d'une particulière gravité reconnue comme incurable au moment du diagnostic" [English translation: "Voluntary in-

terruption of pregnancy may be performed at any gestational age if a physician attests that the pregnancy endangers the woman's health or that there exists a strong probability that the unborn child will be affected by a disease or disability of particular gravity recognized as incurable at the time of diagnosis"] [9]. Ectopia cordis has a very poor prognosis, but survival after surgery has been reported [5]. Prognostic counselling must therefore consider the associated anomalies, diagnostic uncertainty, and available therapeutic options. In this case, surgical repair was unavailable at the treating institution. Whether the condition meets the statutory criterion of a disease "recognized as incurable at the time of diagnosis" requires an individualized clinical assessment within the applicable legal framework [9].

However, the promise of this legal provision remains largely unrealized in practice [9]. Multiple barriers impede its implementation: 1) Low population awareness of the provision; 2) Profound social and religious stigma against abortion [9]; 3) Limited clinical infrastructure for safe abortion provision; 4) Healthcare provider fears of legal consequences despite the legal permissibility of the procedure [9]; 5) Inadequate prenatal diagnosis programmes [6] [8] that would identify fetal lethal anomalies early enough to counsel this option. Consequently, unsafe clandestine abortion remains common, contributing significantly to maternal mortality in Burkina Faso [6].

Institutional surgical limitations and neonatal palliative care

Surgical repair of ectopia cordis was unavailable at the treating institution in this case. This institutional limitation should not be interpreted as evidence that such expertise is absent throughout Burkina Faso or sub-Saharan Africa. Surgical management requires specialized expertise and individualized planning, and may involve staged interventions [10]. Survivors following surgery have been reported, although outcomes depend on the anatomical abnormalities and clinical circumstances [5]. When curative treatment is not feasible, neonatal palliative care can provide dignified end-of-life support, emphasizing comfort, family support, and bereavement counselling [11].

Global context and health inequity

Survival, including long-term survival, has been reported after surgery in specialized centers [5]. Neonatal deaths from congenital anomalies in sub-Saharan Africa remain substantial, a proportion largely preventable through early diagnosis and planned management [12] [13]. Global disparities in maternal and perinatal mortality remain stark [6] [12]. This case starkly illustrates the therapeutic chasm between high- and low-resource settings and underscores the urgent need for equitable access to basic prenatal imaging and care [3] [6]. More broadly, strategies to reduce maternal mortality emphasize effective intrapartum care in primary-level facilities backed by access to referral-level services [14].

5. Systemic Challenges and Recommendations

1) Expansion of obstetric imaging screening [3]: All maternities should have

access to ultrasound equipment and operators trained in second-trimester fetal morphology screening. We propose: a) standardized technician training in fetal structural recognition, b) equipment supply to district maternities, c) telemedicine consultation for suspicious findings. Target: 80% antenatal screening coverage within 24 months.

2) Prenatal fetal anomaly counselling services [8]: Multidisciplinary fetal anomaly clinics should be established to discuss diagnosis, prognosis, and parental options, including legal and ethical considerations regarding pregnancy continuation, termination (where legally permitted) [9], and palliative planning.

3) Neonatal palliative care protocols [11]: Standardized comfort-focused care protocols for neonates with life-limiting conditions, including family support and bereavement services.

4) Clinical team resilience: Establish structured debriefing after difficult cases, training in resilience and grief processing [11].

6. Limitations

This report has significant limitations. First, it is a single case, non-comparative in nature, which limits generalizability across different populations and settings. Second, absence of fetal echocardiography prevented definitive characterization of intracardiac anatomy, a critical limitation in understanding the pathophysiology of congenital heart disease. Third, advanced fetal imaging modalities such as MRI were not performed, which would have clarified diaphragmatic integrity. Fourth, no postmortem examination was conducted due to family preferences, preventing autopsy-based confirmation of structural findings. Consequently, intracardiac anatomy, diaphragmatic integrity, and precise mechanisms of death could not be comprehensively characterized. The presence or absence of associated cardiac structural defects remains unknown and represents an important limitation in defining the true etiology of neonatal death. Additionally, our findings reflect a single institution's experience in sub-Saharan Africa and may not be generalizable to other geographic or socioeconomic settings. Future studies involving larger case series and postmortem investigation would strengthen our understanding of ectopia cordis pathophysiology in resource-limited settings.

7. Conclusion

Complete thoracic ectopia cordis is a rare congenital malformation with a very poor prognosis that varies according to associated anomalies and access to specialized treatment. Survival after surgery has been reported [5], and neonatal death should not be presented as inevitable in all cases. This case illustrates the challenges of late antenatal diagnosis at 37 weeks and the absence of surgical repair capacity at the treating institution. Late detection reduced the time available for parental counselling and multidisciplinary planning, while institutional therapeutic limitations restricted treatment options. These observations do not establish the availability of surgical care throughout Burkina Faso or sub-Saharan Africa.

Priorities include strengthening obstetric ultrasound screening and prenatal counselling networks in rural and semi-urban areas of Burkina Faso. Developing regional referral pathways and access to paediatric cardiothoracic expertise remains an important goal. Clinicians should support timely diagnosis, informed parental counselling regarding legal and ethical options, and individualized neonatal care, including dignified palliative care when curative treatment is not feasible.

Author Contributions

Conceptualization: W.C.L.S., Z.B., D.A.S. Clinical management of the mother and delivery: Z.B., A.S.E.T., S.P.K., J.D.L.C.M., A.D. Prenatal ultrasound diagnosis and imaging: A.S.E.T., S.P.K. Neonatal care and palliative management: F.Z., E.K. Data collection: W.C.L.S., F.Z., E.K. Writing—original draft: W.C.L.S., Z.B. Writing—review and editing: Z.B., A.S.E.T., F.Z., J.D.L.C.M., E.K., A.D. Supervision: D.A.S. All authors read and approved the final manuscript and agree to be accountable for all aspects of the work.

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Ethical Approval

Written informed consent was obtained from the parents. Ethical approval: Health Research Ethics Committee CHU Souro Sanou, Burkina Faso.

Data Availability

Patient data are available from the corresponding author upon reasonable request, under confidentiality agreement.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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