

CASE REPORT

Medical Imaging of Pentalogy of Cantrell: A Case Report from Zambia

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ABSTRACT

Medical imaging plays a significant role in the detection of foetal anomalies, such as the Pentalogy of Cantrell (POC). POC is a congenital anomaly involving five or fewer body malformations: ectopia cordis (EC), tetralogy of fallot (TOF), sternal defects, diaphragmatic defects, and abdominal wall defects. The commonly used imaging modality in the detection of POC during pregnancy is ultrasound because it uses non-ionising radiation, is cheap, and is available in most medical facilities. We report a rare case of POC from a medical imaging standpoint in Zambia. Two obstetric ultrasound scans were performed in the 3rd trimester. The initial scan revealed, among other things, unappreciated abdominal contents, while the second scan revealed a foetal heart sited partially external to the thoracic cavity, with external herniation of the liver and bowels. Our case was classified as incomplete POC because not all five

main malformations were present. This case report highlights the importance of having well-organised obstetric ultrasound imaging services both in urban and rural parts of the country to improve antenatal care.

ABBREVIATIONS

EC	Ectopia Cordis
IUFD	Intrauterine Foetal Death
POC	Pentalogy of Cantrell
TOF	Tetralogy of Fallot
VSD	Ventricular Septal Defect

INTRODUCTION

Pentalogy of Cantrell (POC) is an infrequent cluster of midline inherited birth anomalies that present in the newborn that include defects relating to the heart, pericardium, sternum, diaphragm, and abdominal wall.^{1,2,3} POC may be classified as complete or incomplete.⁴ The complete POC version presents all

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five (5) defects mentioned above, while the incomplete version presents a few congenital anomalies. Literature reports that POC is present in 1 in every 65, 000 births with a regional prevalence of 5.5 per 1 million live births.^{5,6} This foetal anomaly should be diagnosed during pregnancy for appropriate management. Therefore, healthcare professionals involved in the care of pregnant women such as obstetricians, midwives, radiologists, and sonographers should be aware of the condition.

Medical imaging plays a key role in the diagnosis of congenital anomalies. The World Health Organization (WHO)⁷ defines congenital anomalies as structural or functional anomalies that occur during intrauterine life. The main advantage of medical imaging is that it provides a means to appreciate internal anatomy without resorting to invasive means, such as surgery.^{8,9} Medical imaging is used in the diagnosis of birth defects and congenital anomalies, before and after birth. POC is diagnosed before birth using non-ionising imaging modalities, such as Ultrasound and Magnetic Resonance Imaging (MRI).⁸ These two medical imaging modalities are used for pre-birth assessment of foetal anomalies to circumvent the biological effects of ionising radiation on the radiosensitive foetus.^{9,10,11} After birth, ionising radiation medical imaging modalities, such as conventional (plain film) radiography and Computed Tomography (CT), are used to supplement ultrasound and MRI in diagnosing and managing POC.⁸ It is, therefore, important for medical and nursing professionals, such as obstetricians and midwives to understand the advantages and disadvantages of each of these medical imaging modalities in the diagnosis and management of congenital anomalies.

In Zambia and globally, antenatal care (ANC) is one of the approaches employed in healthcare to ensure a woman safely delivers a healthy baby to reduce maternal and neonatal mortalities.^{12,13,14,15,16,17} This includes the diagnosis and management of foetal congenital anomalies. In Zambia, the mortality rates remain considerably high with maternal mortality

estimates at 398 deaths per 100,000 live births.¹⁴ To improve healthcare delivery, the Ministry of Health (MOH) in 2018 produced the ANC guidelines to provide positive pregnancy experiences.¹⁴ Zambia ascribes to the WHO recommendation of 4 antenatal visits for a pregnant woman.^{13,18} During antenatal care, pregnant women are offered antenatal booking, obstetric ultrasound for confirmation of the viability of the pregnancy and gestation age, identification of multiple pregnancies, screening for foetal anomalies, and the placental location.^{19,20} Screening for foetal anomalies, such as POC using ultrasound, is undertaken in the 2nd trimester of pregnancy.^{13,20} In Zambia, obstetric ultrasound scans are predominantly requested by obstetricians and midwives during antenatal visits.

Globally, it is estimated that 295,000 newborns die due to congenital anomalies within 28 days of birth annually.⁷ Radiologists and radiographers/sonographers play an important role in the diagnosis of these congenital anomalies using ultrasound and other medical imaging modalities which improves the management and treatment of these conditions. In this case report, we present a rare case of an incomplete POC from a medical imaging perspective, in Zambia.

CASE PRESENTATION

A 23-year-old female; in her second pregnancy, with one child alive and born at 9 months without abnormalities, was referred from a rural health centre to a general hospital for an ultrasound for suspected mal-presentation. She could not remember her last menstrual period (LMP), therefore her gestational age by dates was unknown and she had no booked ultrasound.

Obstetric ultrasound scan on the first day of admission

On day 1 of admission to the maternity ward of our hospital, the patient was escorted to the department of imaging for obstetric ultrasound by the attending midwife. The ultrasound request form read "obstetric ultrasound to rule out mal-presentation." The sonographer received and welcomed the patient

and the midwife. The patient identification was performed by the sonographer by asking the patient her name, date of birth (DOB), and address. The patient was then informed that an obstetric scan was going to be performed and that she was not going to experience any pain or discomfort.

The patient was Scanned in supine position. A CHISON i3 stationary ultrasound machine with a 3.5MHz curvilinear probe was used in the initial ultrasound scan. The ultrasound findings revealed a single live intrauterine foetus in breech presentation, within adequate liquor volume. The estimated foetal gestation age by a measure of the Bi-parietal Diameter (BPD) was 8.62cm equivalent to 34 weeks 5 Days, with abdominal contents not well appreciated. The ultrasound report was given to the attending midwife. In the maternity ward, the findings were brought to the attention of the patient by the medical officer.

Obstetric ultrasound scan on the second day of admission

On the second day of admission, the midwife noted that the patient went into labour. Patient examination revealed a breech foetal presentation with a foetal heart rate of 136 b/m and regular. On vaginal examination, something protruding from the vulva was felt, perceived to be foetal intestines/bowel. The medical doctor was contacted to review the patient. The medical doctor confirmed that the patient was in established labour, with moderate contractions. The height of the fund us (HoF) was 32/40, breech presentation and regular foetal heart heard. The vaginal examination revealed that the os was 4cm dilated with soft tissue protrusion; not pulsatile, greenish in colour, and perceived as foetal intestine. At this stage, a repeat ultrasound scan was requested.

A Mindray DP10 portable ultrasound machine with a 3.5MHz curvilinear probe was used for the second scan. The ultrasound findings revealed a viable intrauterine foetus with a placenta located posteriorly, but

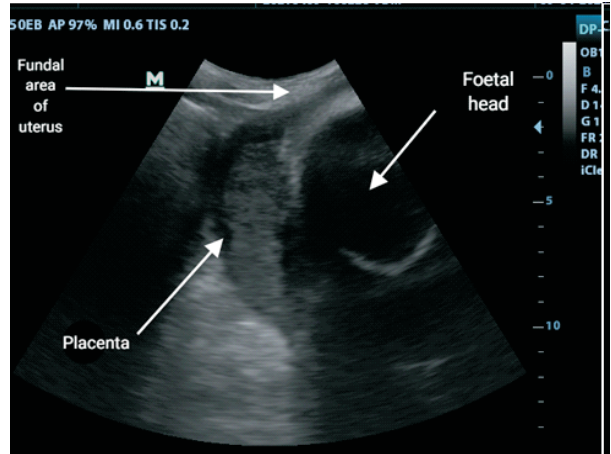


Figure 1: Head and placenta located in the fundal area of the uterus

not low laying. The foetus was in breech presentation, the foetal head and placenta were in the fundal area of the uterus (Figure 1).

Further analysis of the foetus revealed a chest wall defect, with the heart sited partially external to the thoracic cavity (Figure 2a and 2b).

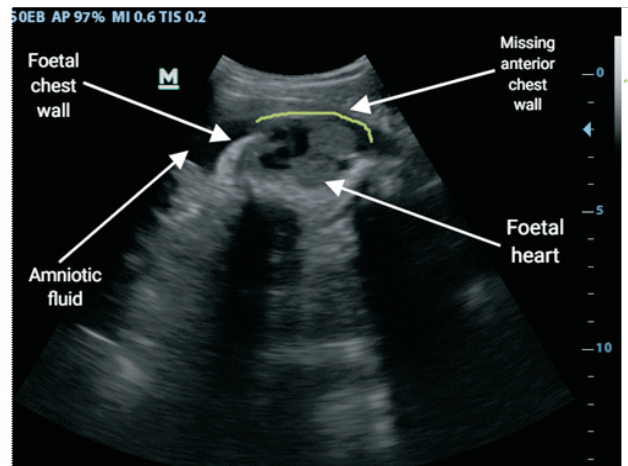


Figure 2a: Visualized foetal heart with open anterior chest wall

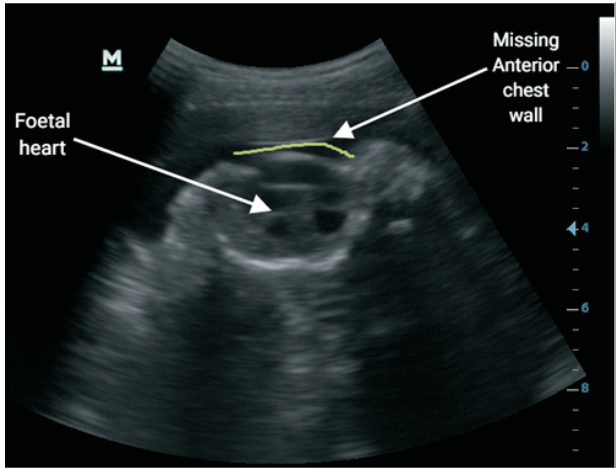


Figure 2b: Visualized foetal heart with open anterior chest wall

Also, a defective anterior abdominal wall was noted, with herniation of the liver and bowels (Figure 3).

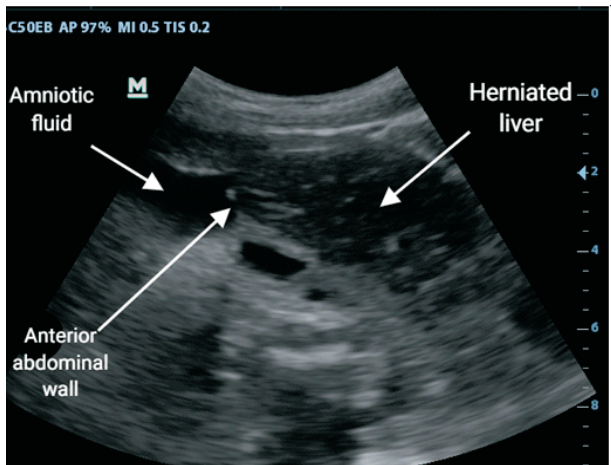


Figure 3: Herniated liver and bowels

The final diagnostic ultrasound report read “single intrauterine foetus, breech presentation with thoraco-abdominal wall defects with Ectopia Cordis and gastroschisis at 32weeks and 6days gestation: a case of Pentalogy of Cantrell”.

The outcome of the pregnancy

The medical doctor noted the ultrasound findings, explained the findings to the patient, and provided counselling. The patient opted for Spontaneous

Vaginal Delivery (SVD) unless all failed, despite being offered CesareanSection. In the late morning, the foetal heart was not appreciated by the midwife and the medical doctor was contacted who confirmed intrauterine foetal death(IUFD). The baby was born by SVD, a female Fresh Still Birth (FSB) weighing 1860g. The baby was noted to have thoraco-abdominal wall defects with EC and herniation of the liver, spleen, and bowels, and poorly formed upper limbs with clenched hands each with two fingers (Figure 4).



Figure 3: Photograph of foetus post-delivery

Pre-delivery and post-delivery counselling were provided to the mother by midwives and the medical doctor. The mother was kept for observation and discharged the following day after it was confirmed that she was fit for discharge.

DISCUSSION

The POC was initially described by Cantrell, Haller, and Ravitch in 1958.²¹ POC may present as complete or incomplete.⁴ The complete version may present five foetal anomalies: EC, TOF, sternal defects, diaphragmatic defects, and abdominal wall defects.⁴ EC is a congenital anomaly where the heart is sited either entirely or partly outside the thoracic cavity. This may be a result of the absence of the sternum also classified under POC. In our case report, the foetus presented with an anterior thoracic wall defect (Figure 2a and 2b), with the foetal heart sited partially outside the chest cavity; hence, EC. TOF is

a congenital heart anomaly that may present in newborns characterized by the presence of pulmonary stenosis, Ventricular Septal Defect (VSD), right ventricular hypertrophy, and overriding of the aorta.^{22,23} On the other hand, incomplete POC does not present with all the five foetal anomalies.⁴ Our case was classified as incomplete POC because the foetus presented only with three features: EC with possible sternal defect, and abdominal wall defect which manifested as external herniation of the liver, spleen, and bowels vis-a-vis gastroschisis. The referral and availability of ultrasound services in our case significantly contributed to the detection of foetal anomalies. However, most of the rural parts of Zambia have no direct access to ultrasound services.²⁴ This is due to a lack of ultrasound machines and trained healthcare professionals.

The diagnosis of POC was made on the 2nd scan. The two scans were done by different sonographers. The initial scan was done by a sonographer with vast experience in the practice yet the anomalies were missed. A sonographer must have a high index of suspicion in such sonographic findings, especially in 2nd trimester when there is lack of appreciation of foetal abdominal content. Various factors can lead to failure to detect foetal anomalies on ultrasound with one of the main ones being the lack of booking systems for ultrasound scans especially for 2nd trimester anomaly scans. Generally, ultrasound scans in many health facilities are not done on a booking basis. All scans: abdominal, small parts, doppler and antenatal scans, among others, are routed through the same radiographer/sonographer in that particular shift, which negatively affects the detection of foetal anomalies. To enhance the detection of foetal anomalies, there may be a need to designate a room for obstetric scans, and book patients in 2nd trimester for anomaly scans. Secondly, obstetric scans for foetal anomaly assessment, in 2nd trimester, must be indicated as such unlike the common indication; routine scans, for foetal viability, gestation age, presentation and so on. Thirdly, and most important, the competence of a sonographer cannot be overemphasized. There

is need for specialized training and other CPD interventions aimed at exposing sonographers to the various types of foetal anomalies and their sonographic appearances.

Various medical imaging modalities, such as ultrasound, MRI, general radiography (plain film), and CT can be used for screening, monitoring, and management of foetal anomalies.^{8,9} However, non-ionising imaging modalities such as ultrasound and MRI are preferred for use prenatally, while ionising imaging modalities, such as general radiography and CT are preferred for postnatal use to protect the unborn foetus from radiation exposure. Ultrasound is the preferred first-line medical imaging method for use in antenatal screening of structural or functional foetal anomalies.^{7,20,25} Other advantages of ultrasound in foetal imaging include less costly, non-invasive, painless, provides high resolution and information in real-time.^{8,19} These highlighted benefits relate well to our case report. Besides, immediately after the woman presented with the part perceived as intestines at the vulva and a provisional diagnosis of gastroschisis made, an obstetric ultrasound scan was urgently requested and performed. However, Kasbanet *al.*⁸ reports that ultrasound is operator-dependent. In the context of this case report, this means having qualified and competent sonographers to perform and detect foetal anomalies. Edwards and Hui,²⁰ adds that enhanced detection of foetal abnormalities via ultrasound imaging is best achieved in dedicated obstetric facilities.

Obstetric ultrasound has a high sensitivity for detecting foetal anomalies such as POC.^{1,2,3,4} Besides, it is recommended that a pregnant woman undergoes an obstetric ultrasound for early detection of foetal anomalies to help them acquire adequate information regarding their condition thereby accessing needed healthcare crucial to their condition and aiding them to make informed decisions regarding whether to keep the pregnancy or terminating it.^{13,20} The findings in our case report confirm the sensitivity of ultrasonography in the detection of foetal anomalies. However, the foetal

anomalies exhibited in our case were detected late in the 3rd trimester. The literature recommends that obstetric ultrasound scans to screen for foetal anomalies are done in the 2nd trimester.²⁰ It has been proven that the foetal anomaly detection rate is higher in the 2nd trimester than in the 1st trimester.²⁶ In the 2nd trimester, the foetal organs are developed which can be detected sonographically.

It should be appreciated that POC may be an accidental finding, especially in a population without regular access to obstetric ultrasound services. Foetal anomalies may also be identified at birth which presents serious emotional trauma to the woman and the family. That said, there is a need to ascertain the level of access to 2nd trimester anomaly scans in Zambia. This data is currently unavailable and may require conducting a study to ascertain the level of access in this regard. The findings of such a study could significantly contribute to sound arguments in favour of increasing the number of ultrasound machines in the country, training and employment of the much needed radiographers/ sonographers to perform obstetric scans. However, it is quite obvious that the findings would reveal substantially low access to ultrasound services as ultrasound services in Zambia are predominantly provided by hospitals which are located in urban areas.²⁴ Antenatal services are classified under primary healthcare and are provided by clinics. Most clinics, where antenatal services are provided, do not offer ultrasound services let alone 2nd trimester anomaly scans. Secondly these clinics are located in rural areas, many kilometres from the nearest hospital providing ultrasound services. The only pregnant women that access ultrasound services would be those that are flagged as having complicated pregnancies and are referred to 1st or 2nd level hospitals for further management.

In cases of POC, echocardiography is used to evaluate defects of the heart vis-a-vis in TOF.²³ Echocardiography provides information on right ventricular hypertrophy if present that results from pulmonary stenosis. In pulmonary stenosis, the heart responds by enhancing right ventricular contractions to push more blood through the

narrowed pulmonary artery which results in increased right ventricular muscle vis-à-vis, right ventricular hypertrophy.^{23,27} Also, Doppler ultrasound can be used to demonstrate regurgitation owing to VSD.²³ Moreover, TOF is identified by foetal echocardiogram in half of the patients during antenatal visits.²³ Our case reports EC as an observed foetal heart-related anomaly and turned out to be a case of Fresh Still Birth (FSB). Therefore, echocardiography was not employed to assess the heart anomalies, such as those classified under TOF. However, in infants with POC that survive delivery, echocardiography significantly contributes to the evaluation of heart anomalies such as TOF, and effective management of the condition even as the infant grows.

In Zambia, there is a shortage of medical imaging professionals, such as radiologists, radiographers, and sonographers in 80% of the facilities.²⁸ These imaging professionals offer imaging services, such as ultrasound. In the context of this case report, the shortage of imaging professionals specialised in ultrasound affects the delivery of obstetric ultrasound services and the detection of foetal anomalies such as POC. The shortage of sonographers is due to a lack of ultrasound study programmes. Currently, only Evelyn Hone College (EHC) in Lusaka province is offering a two-year diploma programme in ultrasound which is not adequate for a population of 18 million. At the time of writing this case report, the University of Zambia (UNZA) in Lusaka had advertised for a master's in ultrasound. The Lusaka Apex Medical University (LAMU) is planning to start offering ultrasound training. The establishment of these study programmes will increase the number of sonographers and improve ultrasound imaging services in the country. This includes the screening and detection of foetal anomalies. In Zambia, ultrasound machines are available in most medical facilities, but MRI scanners are few because of high procurement and maintenance costs.⁸ There are only seven MRI scanners in Zambia: six in Lusaka Province and one on the Copperbelt province. In this case report, MRI was not performed because it is not available at our hospital and in the province.

CONCLUSION

This case report highlights the importance of well-organised antenatal care services supported by ultrasound to identify and treat conditions that may threaten the health of the foetus and the mother. This includes the early identification of foetal anomalies for appropriate management and counselling of pregnant women. The Ministry of Health (MOH) should ensure that pregnant women in both urban and rural parts of Zambia have direct access to obstetric ultrasound imaging services. This can be achieved by the procurement of more ultrasound machines and recruitment of radiographers/sonographers including in-service training of radiographers in advanced ultrasound practice. Most importantly, the screening of foetal abnormalities can be best provided through dedicated obstetric facilities in each of the ten provinces of Zambia.

CONSENT

Permission to use images of the foetus and access the patient's notes was obtained from the Medical Superintendent who is the controlling officer. Also, an ethical waiver was obtained from the Lusaka Apex Medical University Research Ethics Committee (reference number 00425-21).

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