International Medical Case Reports Journal

Dovepress
Taylor & Francis Group

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CASE SERIES

Cyclopia, A Rare and Lethal Congenital Anomaly: Report of Two Cases, 2025

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Background: Cyclopia is an extremely rare and fatal congenital anomaly in which causes are not commonly known. This condition is marked by the failure of the eye orbits to properly divide, resulting in a single eye field or closely positioned eye fields. There is no specific treatment, and the prognosis is generally poor with most babies dying shortly after birth or during infancy.

Case Presentation: These two newborns were born to multiparous mothers, which died soon after birth. Diagnosis was made postpartum with centrally located single eye and long proboscis on the midline of the forehead, with the second had associated with cystic hygroma. Both cases had no other gross visible congenital anomalies on the extremities, abdomen, and head. But other anomalies as renal, central nervous system, cardiac and chromosomal anomalies cannot be excluded.

Conclusion: Both cases had died immediately, and health professionals should experience with detailed antenatal ultrasound scanning to detect these congenital anomalies antepartum for better counseling of patients. Routine antenatal ultrasound assessment is crucial in low resource countries where ultrasound scanning is commonly done in higher health facilities.

Keywords: cyclopia, holoprosencephaly, congenital anomaly, Eritrea

Introduction

Cyclopia is a rare and severe form of holoprosencephaly, a condition in which the forebrain fails to divide into two hemispheres during embryonic development,[1–8] encountered in approximately 1 in 100,000 births.[5] Cyclopia typically presents with a median single eye or a partially divided eye in a single orbit, absent nose, and a proboscis above the eye.[8] This condition is marked by the failure of the eye orbits to properly divide, resulting in a single eye field or closely positioned eye fields. Additional features include a missing or dysfunctional nose and malformed ears and can occur in isolation or as part of a broader genetic syndrome.[2,3]

The etiology of this rare syndrome is still largely unknown and most cases are sporadic; however, heterogeneous risk factors, including genetic factors and chromosomal anomalies, have been implicated as possible causes.[8,9] Cyclopia leads to a stillbirth since the brain and other parts of the body do not grow normally in fetuses with this disorder.[6] Cyclopia is a malformation incompatible with life[2,8] and most embryos with this condition are aborted, stillborn, or die shortly after birth.[2,3,6] There is no specific treatment, and the prognosis is generally poor with most babies dying shortly after birth or during infancy.[1] The prenatal diagnosis of cyclopia can be made early by ultrasound, and the awareness of the spectrum of sonographic findings of cyclopia can improve the accuracy of prenatal diagnosis.[8,10] Antenatal ultrasound should be performed to lead to early detection of such rare cases, which are incompatible with life, and termination of pregnancy should follow.[11]

Early diagnosis during pregnancy and proper management should be emphasized to prevent further psychological harm to the mother with this syndrome.[6] In addition to the adverse medical outcomes, the broader socio-cultural implications, psychological effects, and interplay with spirituality cannot and should not be ignored in resource-limited settings in sub-Saharan Africa.[1] Thus, the need for heightened antenatal surveillance and mass education to

https://doi.org/10.2147/IMCRJ.S567352
Received: 13 October 2025
Accepted: 16 January 2026
Published: 2 February 2026

demystify the occurrence of congenital anomalies is imperative.[1] To reduce such incidences, more awareness and emphasis must be made out on early ultrasound diagnosis of fetal anomalies and regular antenatal visits.[7]

It is poorly reported in sub-Saharan Africa.[1–3] In developing countries, many cases remain undiagnosed due to the lack of regular prenatal care.[3,9] In this case report, multiple unusual abdominal defects were present, including a huge omphalocele containing whole liver and spleen, urinary bladder extrophy, and undefined abnormal external genitalia.[8] This rare condition is further reported poorly with knowledge gap in diagnosing with ultrasound. Their epidemiology is not clearly determined; thus, we will report two cases of cyclopia clinical significance to increase knowledge of health professionals in diagnosing such cases.

Case Reports

Case 1

Maternal History and Antenatal Findings

The first case was born to a multiparous mother with previous normal delivery presented in labor to the delivery ward. She had two times ANC follow-up in nearby health facility, and anatomic ultrasound scanning was not done by experienced health personnel in higher health facility. She did not have any chronic illness or medication intake or had no similar problem previously.

Intrapartum Events

During her arrival to Orotta National Referral Maternity Hospital, she was in the active phase of labor and labor progressed smoothy. By the time she reached 8cm cervical dilatation, there was persistent fetal bradycardia despite resuscitation, and an emergency cesarean section was done for fetal bradycardia.

Neonatal Findings and Outcome

A 3.5kg neonate with Apgar score of 3 and 4 in the first and fifth minute was delivered, which died immediately. The newborn had centrally located single eye, with tubular nose-like structure above the orbit, without other gross visible congenital anomalies on the extremities, abdomen, neck and head. But other anomalies as renal, central nervous system, cardiac and chromosomal anomalies cannot be excluded (Figure 1).

Case 2

Maternal History and Antenatal Findings

The second case was a 24-year-old gravida 3 para 2 mother with gestational age of 34 weeks presented with labor pain on 23/02/2025 to Orotta National Referral Maternity Hospital. She had poor antenatal follow-ups and had no history of any medication intake or chronic diseases.

Intrapartum Events

During admission, she was in labor, and ultrasound revealed a single viable fetus with breech and cystic hygroma (Figure 2). Emergency cesarean section was done for persistent fetal bradycardia.

Neonatal Findings and Outcome

A 2.5kg female with poor Apgar score was delivered. Neonate had multiple anomalies with centrally located eyes, protrusion in the forehead. (Figure 3) A large cystic hygroma of about 6 x 8cm was clearly noted from the posterior neck (Figure 4) without visible other anomalies on the other body parts. (Figure 5) The neonate died immediately after birth and the patient was counselled before and after delivery.

This data was collected by senior residents in the department of obstetrics and Gynecology during routine obstetric care on delivery. This was carried out in Orotta National referral Maternity Hospital, situated in Asmara, the capital city of Eritrea. Both patients (mothers) gave written informed consent to publish this data for educational purpose, but ethical approval statement was not sought for these case reports as it is not applicable. Other congenital anomalies were not scanned by ultrasound and postmortem exam was not done for both cases. Table 1 shows comparison among different paraments between the two cases.

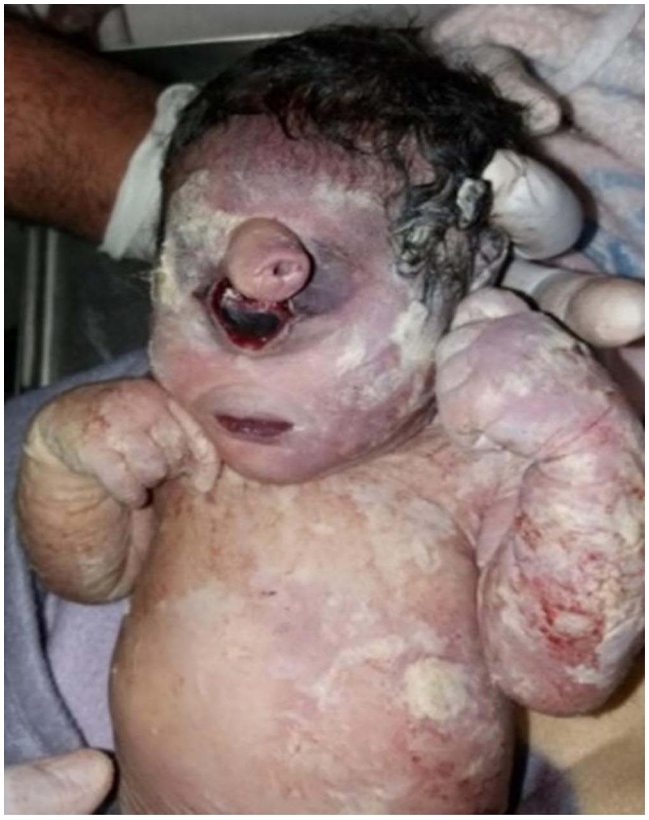

Figure 1 Cyclopia of the first case with centrally located single eye and tubular nose-like structure above the orbit.

Discussion

Cyclopia is a rare and lethal congenital anomaly, which could be associated with various malformations. The second case had associated cystic hygroma. Similarly, one case report indicated that cyclopia is frequently accompanied by other malformations.[5,7] Besides, studies reported that unusual abdominal defects, including a huge omphalocele, urinary bladder extrophy, and undefined abnormal external genitalia.[8] Thus, even though cyclopia is fatal, evaluating for other associated congenital anomalies is essential. Holoprosencephaly is violation of the normal telencephalic splitting of the forebrain into bilateral hemispheres of the brain resulting from incomplete splitting of the prosencephalon into the right and left hemispheres. It occurs between 18 and 28 days of pregnancy.[10,11] The etiology of this rare syndrome is still largely unknown, but heterogeneous risk factors as chromosomal anomalies have been implicated as possible causes.[8,9] If the testing was available for chromosomal anomalies in these cases, specific cases such as trisomy 13 could be associated with cyclopia.

The clinical presentation of the newborns in both cases was with a single central eye, absence of nose, and proboscis above the eye. Different case reports indicated similar results with a single central orbital fossa with a tubular nose-like appendage above the orbit.[1–7,9] Knowing this clinical presentation for health professionals is very crucial for early diagnosis and family counseling during the antenatal period. Both these cases had no identifiable risk factor. Similarly, literatures reported that the cause is not known in most cases.[8,9]

Cyclopia has a fatal outcome with most aborted and most of those reached viability delivered stillbirth or died immediately as in both these cases. This was consistent with many literatures that the newborn was delivered a stillborn[5,7,9] and died immediately after few minutes.[1–4,6] Early perinatal diagnosis and determination of severity are important to inform parents of their baby's life chances.[4]

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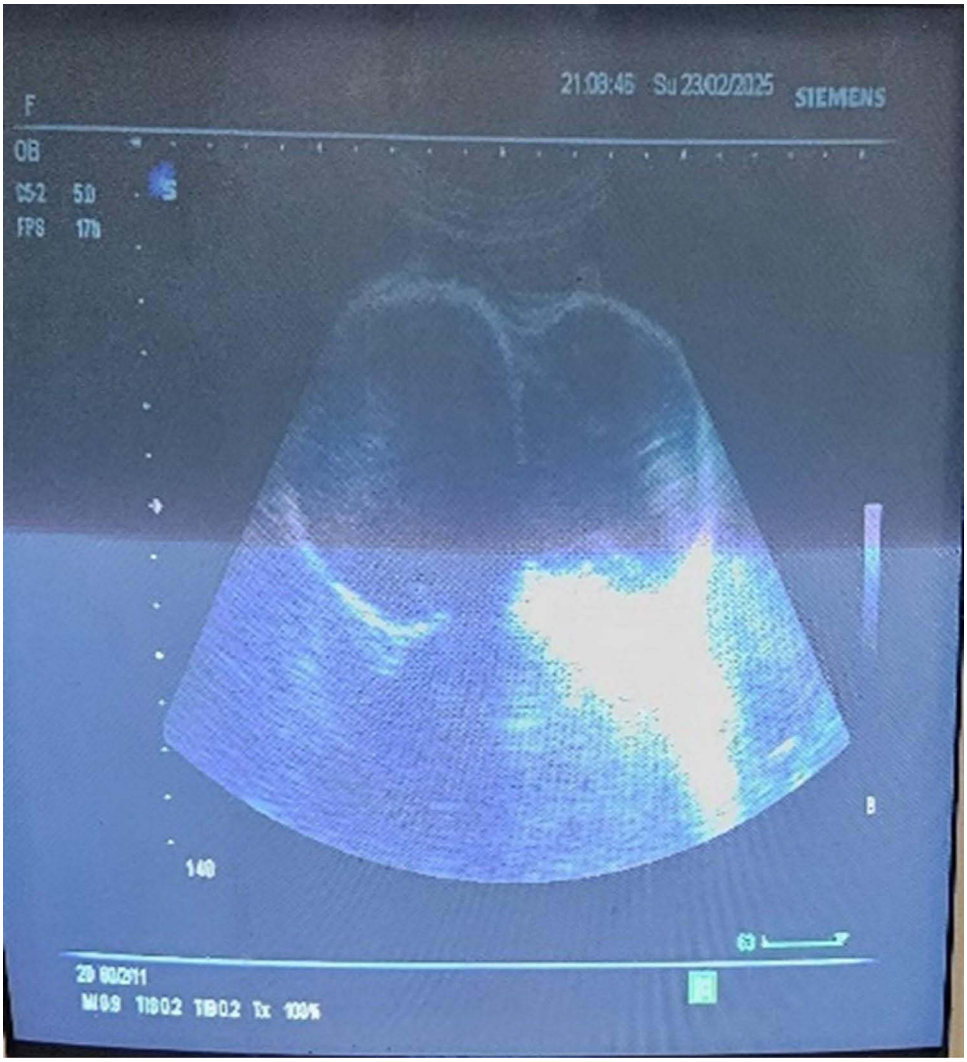

Figure 2 Transabdominal ultrasound of the second case with single viable fetus, and breech presentation complicated with cystic hygroma.

In developing countries where the antenatal care is poor or, if patients are following in lower health facilities, the abnormality may not be detected antenatally. Similarly, these cases were following their ANC in lower health facility and diagnosis was made postpartum. Other studies reported that in developing countries, where pregnant women often lack regular prenatal care, many cases go undiagnosed.[3] With advances in fetal imaging and availability of high-resolution ultrasound machines, it is surprising that such lethal malformations are still presenting after the period of viability.[7]

Due to its rarity, health physicians should have high index of suspicion during antenatal ultrasound screening. The awareness of the spectrum of cranial, facial and extra-facial sonographic findings of cyclopia can further improve the accuracy of prenatal diagnosis.[9] All antenatal units, therefore, should incorporate routine antenatal anomaly ultrasound in their program to detect such abnormalities.[10] With the advancement in antenatal USG to identify malformed fetuses early in pregnancy, the incidence of such cases progressing to advanced gestations is decreasing.[9]

If patients have this fatal congenital anomaly, they should be counseled for their current and next pregnancy. Literature indicates that raising awareness among parents about the importance of monitoring the pregnancy, and the interest in psychological support for parents, especially young people, in the event of a rare malformation incompatible with life.[2] It is therefore important to inform parents/families of the abnormality and the possible outcome during the antenatal period, decide on whether termination of pregnancy may be undertaken.[10]

Figure 3 Cyclopia of the second case with centrally located eyes, protrusion in the forehead.

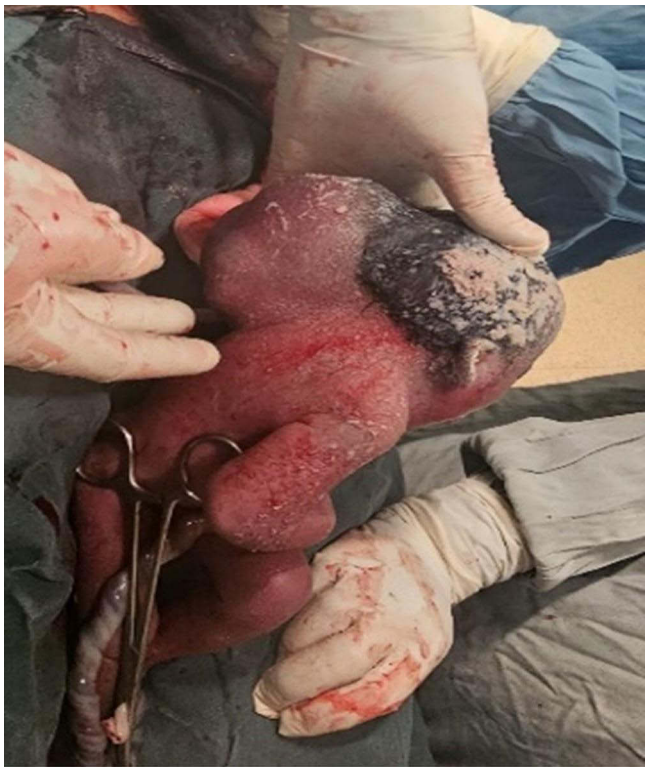

Figure 4 Big cystic hygroma of about 6x8cm of the second case.

https://doi.org/10.2147/IMCRJ.S567352

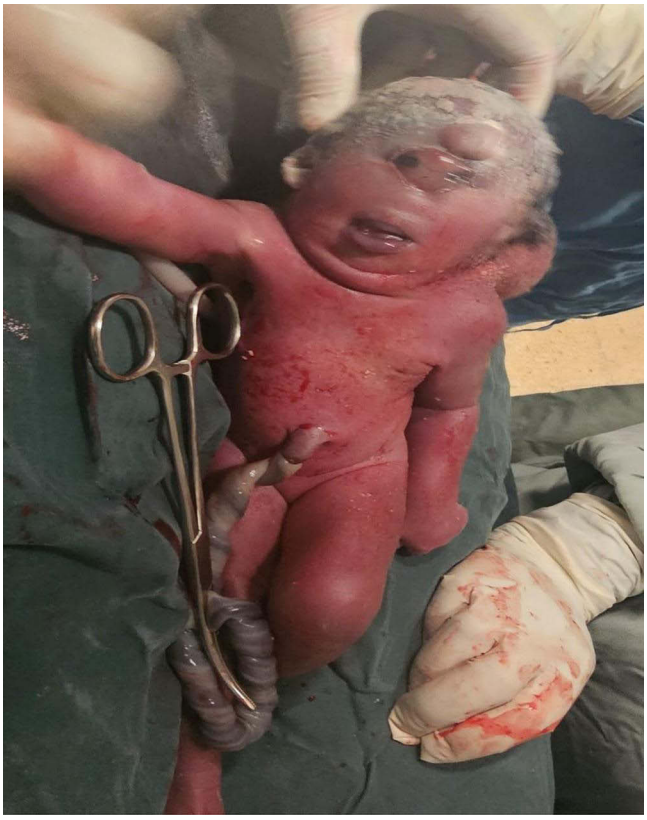

Figure 5 Cyclopia with other body parts as abdomen and extremities.

These reports highlight the significance of antenatal ultrasound scanning by experts to diagnose these lethal anomalies which implicates for policy and practice in health systems in Eritrea. This case report has limitations as absence of genetic testing, inability to perform fetal MRI, lack of autopsy to reach further definitive diagnosis and associated congenital anomalies.

Table 1 Comparison Among Different Paraments Between the Cases

Parameters/Variables	Case 1	Case 2
Gravidity and parity	Multigravida and multiparous	Multigravida and multiparous
Gestational age on visit	Term	Preterm, 34weeks
Antenatal care utilization	Poor (two times) at lower facility	Poor (two times) at lower facility
Diagnosis for cyclopia	Intraoperative (postpartum)	Intraoperative (postpartum)
Mode of delivery	Cesarean section, 3.5kg poor Apgar	Cesarean section, 2.5kg poor Apgar
Clinical findings	Centrally located single eye, with tubular nose-like structure above orbit	Centrally located eyes, protrusion in the forehead and big cystic hygroma
Outcome	Immediate death	Immediate death
Associated visible anomalies	No visible anomalies	Yes, cystic hygroma

Conclusion

This extremely rare and associated cystic hygroma in one case had immediately died after birth. Health professionals should have high index of suspicion and exercise antenatal ultrasound to detect this anomaly and plan for management.

Consent

The guardians of both patients gave written consent for the publication of the case details and accompanying images, but institutional approval was not sought for case reports and was not applicable.

Acknowledgment

The authors acknowledge all hospital staff for their inpatient care and follow up.

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.

Funding

This case report had no source of funding.

Disclosure

The authors have no conflicts of interest to disclose for this work.

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