

"Would This Happen Again?" Addressing Recurrence Risks in Cyclopia: A Case Report

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Case Report

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Abstract

Background

Cyclopia is a severe, rare congenital anomaly characterized by the failure of the orbits to divide, resulting in a single midline ocular structure positioned beneath a rudimentary nasal prominence (proboscis). This case highlights the profound psychological impact on the family when the diagnosis is unexpected and missed during antenatal care (ANC). Timely prenatal detection creates crucial opportunities to prepare parents emotionally, allow healthcare providers to address concerns about recurrence risk in subsequent pregnancies, and deliver compassionate, supportive care.

Case Presentation:

A 21-year-old primigravida at nine months of gestation presented in active labor and delivered a 3,600-gram female neonate exhibiting severe dysmorphic features consistent with cyclopia, including a single central eye and an absent nose. The immediate postpartum period was marked by an acute stress reaction characterized by emotional numbing and flat affect. The care providers, unfamiliar with the rare condition, lacking training in perinatal palliative care and grief counseling, and constrained by time for immediate consultation, were unable to address key questions from the family under their care.

Conclusion

The birth of an infant with cyclopia is a devastating experience, particularly when left undiagnosed until delivery due to lack of awareness or underutilization of routine antenatal care (ANC). Strengthening community awareness regarding ANC and equipping healthcare providers with specialized training can improve early recognition, allowing families to receive timely guidance and compassionate support.

Introduction

Cyclopia is one of the most profound and rare congenital anomalies in human development, characterized by the failure of the orbits to form independently, resulting in a single eye structure centrally positioned beneath a rudimentary nasal prominence. Occurring in approximately 1 in 100,000 live births, this condition is the most severe phenotypic manifestation of holoprosencephaly (HPE), a developmental failure wherein the primitive forebrain fails to undergo proper cleavage into bilateral hemispheres [1–3].

This case report highlights the profound psychological consequences that arise when severe congenital anomalies, particularly those with gross dysmorphic features such as cyclopia, remain undiagnosed in the absence of routine antenatal care (ANC). In rural settings, where such anomalies may carry significant cultural meaning, the lack of prenatal detection leaves families entirely unprepared for the

visual and emotional shock of delivery. In this patient, the undetected development of cyclopia transformed what should have been a routine milestone into an acute traumatic event. Crucially, this report also exposes a significant deficit in frontline clinical preparedness. The attending healthcare providers, themselves profoundly shocked, were unable to offer evidence-based explanations or structured psychological support. Their response was limited to basic comfort and the dispelling of cultural stigma, merely reassuring the family that the condition was not a curse, without the capacity to articulate its medical etiology. This case therefore serves as a critical call to action, demonstrating the urgent need to expand basic antenatal diagnostic coverage in rural regions and to equip primary healthcare workers with training in both the clinical recognition of rare congenital anomalies and the delivery of compassionate, evidence-based grief counseling.

Case Presentation

A 21-year-old primigravida at nine months of amenorrhea presented in active labor. She delivered a 3,600-gram live female neonate with a gross facial deformity characterized by a single centrally located eye and absence of a fully formed nose (Fig. 1). The neonate had an Apgar score of 8 at birth but exhibited signs of life for only 20 minutes.

The mother was visibly shocked by the unexpected outcome and exhibited an acute stress reaction, initially refusing to speak for several hours despite emotional support and reassurance from the attending midwife and general practitioner. When she eventually spoke, her primary concern was whether the anomaly would recur in a future pregnancy. However, the attending care providers, unfamiliar with the rare condition, lacking training in perinatal palliative care and grief counseling, and constrained by time for immediate consultation, could offer only vague reassurance rather than an evidence-based answer regarding holoprosencephaly and its genetic implications.

Subsequently, a community elder among the accompanying family members provided culturally appropriate counseling, which appeared to help ease her grief. The mother later accepted the situation and was discharged in stable clinical condition with no expressed or observed suicidal ideation. She was counseled on the importance of routine antenatal care follow-up for subsequent pregnancies, though formal genetic counseling was unavailable at the facility.

Discussion

During normal early embryogenesis, the cranial neural tube undergoes rapid expansion into the prosencephalon, mesencephalon, and rhombencephalon. By the fourth and fifth weeks, the prosencephalon must undergo "ventral induction" it is a process of midline cleavage that divides it into the telencephalon and diencephalon. Simultaneously, the eye field, initially a single region in the anterior neural plate, relies on the signaling molecule Sonic Hedgehog (SHH) to split into two lateralized primordia. This bifurcation is the critical event that ensures the development of bilateral, separate optic

vesicles [4]. Cyclopia occurs when this sequence is disrupted, resulting in a severe form of holoprosencephaly.

The etiology of cyclopia is multifaceted, arising from a complex interplay of chromosomal abnormalities, genetic mutations, and environmental teratogenic factors. Chromosomal aberrations are frequently implicated, with Trisomy 13 (Patau syndrome) being the most common disorder associated with holoprosencephaly (HPE). Other, less frequent associations include Trisomies 18 and 21, as well as instances of triploidy. Because this developmental failure occurs during the earliest stages of neurulation, the process by which the neural tube is formed, the resulting structural anomalies are profound and almost universally incompatible with postnatal life [1–3].

The recurrence risk for cyclopia, the most severe form of holoprosencephaly (HPE), is highly dependent on its underlying etiology. In chromosomally normal fetuses with HPE, the recurrence risk is reported to be 6%, but this drops to 1% in cases with an abnormal karyotype. For monogenic forms, the risk is much higher, with up to 50% recurrence risk for autosomal dominant traits and 25% for autosomal recessive traits [5].

More specifically, in families with autosomal dominant HPE, the risk for an obligate carrier to have a child with severe HPE is estimated to be 16–21%, with an overall risk of 29–35% for any manifestation of the disorder. Factors associated with recurrence include a positive family history of HPE or its microforms (such as a single maxillary central incisor or cleft lip), and parental consanguinity, which suggests an autosomal recessive inheritance pattern. Advanced maternal age is a risk factor for chromosomal anomalies like trisomy 13, which can present with cyclopia, while other factors such as maternal diabetes and in vitro fertilization (IVF) have also been implicated in some cases [6, 7].

First-trimester ultrasonography remains an effective method for the early detection of cyclopia [8]. However, in resource-limited settings the lack of standardized, accessible prenatal care remains a significant barrier (mainly lack of awareness among pregnant women). Consequently, many affected pregnancies remain undiagnosed until birth, placing an unexpected and severe psychological burden on families who have not been prepared for the clinical reality of the neonate's condition [9, 10].

In Ethiopia, maternal health services have made notable progress in overall contact rates, yet a significant drop-off persists when evaluating complete adherence and quality of care. Analysis linking national household and facility datasets reveals that while nearly two-thirds of expectant mothers engage in at least one primary healthcare visit, only about one-third complete the minimum standard of four encounters, and adjusting for facility readiness and evidence-based process quality reduces effective antenatal coverage to just 12.3% nationwide [11]. This widespread systemic drop-off is heavily driven by entrenched socioeconomic disparities, including low maternal literacy, pastoral or rural residence, and geographical distance to clinical infrastructure [12, 13].

These coverage gaps carry severe psychological and operational consequences when critical health conditions go unmonitored during gestation. Unexpected birth-room discoveries of severe structural

malformations generate immense emotional and clinical strain for families and delivery teams, transforming what should be a milestone into a period of profound shock. When major physical anomalies remain unidentified prior to labor, clinical providers lose the critical window to deliver empathetic non-directive counseling, coordinate specialized intrapartum protocols, or prepare palliative measures beforehand. Under national clinical practice standards (Ethiopian Ministry of Health, 2021) [14], baseline antenatal care protocols mandate a routine baseline sonogram before 24 weeks' gestation for all expectant mothers to establish gestational age and identify major physical malformations. However, resource limitations, equipment shortages, and diagnostic skill gaps across lower-tier facilities mean that mothers without recognized high-risk indicators rarely receive timely, high-quality imaging [15]. Bridging the gap between primary contacts and true, quality-adjusted antenatal care thus requires not only boosting continuous visit completion, but also strengthening baseline diagnostic capacity so that families receive compassionate, comprehensive, and proactive care throughout pregnancy.

Limitations

This case report has several limitations. First, genetic testing was not performed because of the limited availability of genetic diagnostic services and resources in our setting; therefore, the underlying genetic basis could not be determined. Second, the mother was not followed through subsequent pregnancies, and consequently, we were unable to assess the recurrence risk or document pregnancy outcomes in future pregnancies. These limitations highlight the need for improved access to genetic testing and long-term follow-up and counseling for families affected by cyclopia.

Conclusion

The birth of an infant with cyclopia is an overwhelming and isolating experience, particularly when left undiagnosed prior to delivery. When prenatal diagnosis is missed, the delivery room becomes the primary space for acute crisis intervention; therefore, healthcare systems must equip frontline care providers to deliver sensitive, culturally appropriate bereavement support. By prioritizing human dignity, reproductive health education, and provider readiness, we can ensure no family is left to navigate the sudden, profound loss of a child without answers or support.

Abbreviations

ANC

Antenatal care

HPE

Holoprosencephaly

SHH

Sonic Hedgehog

Declarations

Ethical Approval and Consent to participate: NA

Consent for publication: Written informed consent was obtained from the patient for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

Availability of Data and Materials: No, we do not have any research data outside the submitted manuscript file.

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Author's contribution

GKK: Conceptualization, data curation, literature search, investigation, formal analysis, writing–original draft

STM: Patient Care, data curation, literature search, formal analysis, writing–review and editing.

KTH, BMW, EFY: data curation, literature search, formal analysis, writing–review and editing.

BTH: Patient Care, Conceptualization, data curation, literature search, investigation, formal analysis, writing–original draft.

All authors approved the final version and agree to be accountable for all aspects of the work.

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Figures



Figure 1

Clinical photograph of a newborn with cyclopia, characterized by a single diamond-shaped central ocular structure and absence of nasal development, alongside diffuse post-delivery vernix caseosa coating the head, trunk, and extremities.

