



## CASE REPORT

# Cyclopia, A Rare Congenital Malformation: A Case Report in a Resource-Limited Setting in Sub-Saharan Africa

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## Abstract

Cyclopia is a rare and severe form of holoprosencephaly, a condition in which the forebrain fails to divide into two hemispheres during embryonic development. It is poorly reported in sub-Saharan Africa. We report the case of a 21-years-old G2P1001 lady who was referred to our service for better management of preterm labor. She was 29 weeks pregnant, and her cervix was 4 cm dilated on arrival. Labor progressed to vaginal delivery of a preterm malformed baby girl. Physical examination of the neonate revealed; a preterm gasping female baby, weighing 1.26 Kg, Apgar score at birth was 5/10, there was a single midline eye with two distinct irises, a 2 cm long proboscis on the midline of the forehead, and the absence of a nose. The heart sounds were irregularly irregular. The baby died 5 minutes after delivery. We reviewed the sociocultural implications in this resource-limited setting as the father of the baby abandoned the mother of the baby in the hospital as he associated the congenital abnormality of the baby to witchcraft.

## Keywords

Cyclopia, Holoprosencephaly, Congenital malformation, Resource-limited setting, Cameroon

## Introduction

Holoprosencephaly (HPE) is a complex brain malformation resulting from incomplete separation of the prosencephalon, occurring between the 18<sup>th</sup> and the 28<sup>th</sup> day of gestation and affecting both the forebrain and the face. It is estimated to occur in about 1/16,000 live births and 1/250 pregnancies [1]. Cyclopia with

a proboscis is even more rare (1/100,000 newborns and stillbirths) and accounts for the most severe form of holoprosencephaly. It generally occurs due to the incomplete separation of the prosencephalon into the two halves of cerebral hemispheres during organogenesis [2,3]. We herein report a case of cyclopia with proboscis occurring in a low- and medium-income country (LMIC) in sub-Saharan Africa, and review the literature.

## Case Description

This is the case of a 21-years-old G2P1001 lady pregnant lady who gave birth to a 29-weeks-old female neonate through the vagina at the Tubah District Hospital, Northwest Region, Cameroon. She had a previous vaginal delivery of a normal, live female baby at term three years ago, who is apparently in good health. She is a cook and has a first-degree family history of diabetes and hypertension. For the current pregnancy, she started antenatal clinic at 22 weeks of gestation in the Bambili medicalized health center. She attended twice in total. The laboratory results at the first visit were as follows; blood group = B Rhesus positive, HIV= negative, urinalysis = normal, VDRL = positive, TPHA = negative, toxoplasmosis antibodies = negative, hepatitis B surface antigen = negative, vaginal smear = normal, and hemoglobin level = 10.5 g/dl. Obstetric sonography during the booking visit revealed mild

ventriculomegaly, which was not given further medical attention. There was no history of alcohol consumption throughout the pregnancy, and the patient denied taking any drugs before her booking visit. Also, the patient denied a family history of congenital malformations and consanguineous marriages. There was no known exposure to teratogens. At the booking visit, she was prescribed the following medications, which she took accordingly; iron and folic acid, pyrimethamine and sulfadoxine, mebendazole, calcium tablets, and tetanus toxoid.

She was referred from Bambili medicalized health center to our service for better management of preterm labor and premature rupture of membranes at 29 weeks gestation. On admission, she complained of gush of fluid per vagina of 8 hours duration and labor-like pains of 6 hours duration. Physical examination revealed a well-looking lady with stable vital signs; the cervix was 5 cm dilated, centralized, and 80% effaced, no membranes were felt, the presentation was cephalic, fetal descent was 2/5, and the fetal heart rate was 134 bpm. The patient was administered ampicillin 2g as antibiotic prophylaxis for premature rupture of membranes and dexamethasone 8 mg intravenously to facilitate lung maturity. Labor progressed and 2 hrs later, she delivered a female preterm baby per vagina. Her birth weight was 1.26 kg, her length was 41 cm, and head circumference of 30 cm. She had a single midline eye with two distinct irises, a 2 cm proboscis on the midline of the forehead, and the absence of a nose (Figure 1). The mouth and

outer ears were normal. There were no additional digits, spine deformities, or umbilical malformations observed. The newborn gasped to death 5 minutes after delivery. The third stage of labor and the postpartum period for the were uneventful.

Mother and family were given proper counseling on congenital malformations and the possibility of having normal children in subsequent pregnancies. Despite optimal counseling of the couple, the husband abandoned her about 2 hours after delivery, associating the congenital malformation to witchcraft. The mother was discharged on the third day after delivery and was reviewed 1 week late with no complaints.

## Discussion

To the best of our knowledge, this is the first reported case of cyclopia in Cameroon. Holoprosencephaly is a brain malformation that often presents with facial anomalies such as closed ocular orbits, microcephaly, cleft lip, and cleft palate. It results from the failure of the prosencephalon to adequately develop and separate into left and right cerebral hemispheres [4-8]. Holoprosencephaly is divided into three types, including lobar, semi-lobar, and alobar (cyclopia) holoprosencephaly. Worldwide, cyclopia with proboscis is the rarest and most lethal form of HPE and is incompatible with extrauterine life as was observed in our case [3,9-11]. In cyclopia, the brain does not divide, with a resultant undifferentiated cerebral hemisphere associated with a monoventricles, cranial midline



**Figure 1:** The newborn with cyclopia.

abnormalities, and fusion of the thalamus [3,9,10]. Some syndromic sequences that have been associated with cyclopia include Trisomy 13, Pseudo-trisomy 13, Smith-Lemli-Opitz Syndrome, Pallister-Hall Syndrome, and other Aneuploidies [9]. Clinically, cyclopia often presents with severe facial defects, usually a single eye field and a proboscis above the eye [10-12]. Some non-facial features include polydactyly, ventricular septal defects, renal dysplasia, and omphalocele, which often depicts poor prognosis and most often presents with stillbirth [2,9,10,12]. In this case, we found the classical facial features of cyclopia, including a single median orbit, micrognathia, absence of the nose, and a proboscis above the eye. However, we could not exclude extracranial anomalies.

The majority of holoprosencephaly cases are sporadic and the exact cause remains unknown [13]. However, various heterogeneous factors, mainly genetic and environmental, have been implicated. These include chromosomal abnormalities (chromosomes 3 and 10), alcohol consumption, toxoplasmosis, rubella, cytomegalovirus and herpes simplex (TORCH) infections, maternal diabetes, maternal exposure to teratogenic drugs (lithium, aspirin, retinoic acid, amidopyrine, corticosteroids, anti-convulsant) and ionic radiation [11,14,15]. In our case, none of these factors were identified, although we cannot completely rule them out since the use of over-the-counter drugs and street medications are common in our setting [16,17]. Antenatal ultrasonography is very effective in the diagnosis of holoprosencephaly between 21 weeks and 35 weeks of gestation and a confirmation is made by a coronal view of the brain in the presence of the absence of cavum septum pellucidum with squaring and fusion of fontal horns. This malformation most often manifests with ventriculomegaly as was seen in this case; therefore, HPE should be assessed in all cases of fetal ventriculomegaly [18]. Magnetic resonance imaging of the fetal brain can be done in cases where sonographic findings are inconclusive [19,20]. In this case, ventriculomegaly was reported by the radiologist technician at 22 weeks- and 29 weeks gestation. However, no further investigation was done, probably due to limited expertise in this resource-limited setting. The management of cyclopia generally involves the termination of pregnancy after a thorough prenatal assessment.

Overall, congenital malformations are a global health issue affecting many countries; however, the acceptability rate of these defects and their aetiologies varies among different cultures. In some settings, especially in LMICs, it is often seen as a taboo with deleterious consequences [21,22]. As noted in our case, the resultant altered perception, stigma, and discrimination associated with this condition could break family bonds, lead to social exclusion, and further

widen the gap to access healthcare. This case report is aimed at increasing awareness of the occurrence of cyclopia and the need for a high level of suspicion in all fetuses with abnormal intracranial sonographic findings such as ventriculomegaly. This will enable effective counseling and planning of care.

## Conclusion

Cyclopia with proboscis is a rare and severe form of holoprosencephaly and can occur in isolation or as part of a broader genetic syndrome. There is no specific treatment, and the prognosis is generally poor with most babies dying shortly after birth or during infancy. 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. Thus, the need for heightened antenatal surveillance and mass education to demystify the occurrence of congenital anomalies is imperative.

## Conflict of Interest

None.

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