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Case Report

Cyclopia: A rare case of congenital anomaly associated with holoprosencephaly

Nalmiades Ambra[1]*

[1]*Community Health Center, Lingga, Sungai Ambawang Sub-district, Kubu Raya Regency, West Kalimantan, Indonesia*

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ABSTRACT

Cyclopia is a rare congenital disorder characterized by the fusion of the eyes, typically as part of holoprosencephaly, resulting from a failure of the forebrain to divide during fetal development. Occurring in approximately 1 in 13,000 to 20,000 births, many cases go undetected due to early miscarriages. The condition is associated with significant facial deformities, including an absent or severely underdeveloped nose and malformed ears. This case report details a 31-year-old woman who presented at 27 weeks of gestation with abdominal pain and bleeding. Ultrasound revealed a single-lobed brain, hypotelorism, and cyclopia in the fetus. A vaginal delivery was planned, resulting in the birth of an 800-gram baby boy with severe facial anomalies who died shortly after birth. Holoprosencephaly encompasses varying degrees of brain malformation, with cyclopia being the most severe form. The absence of effective treatments necessitates early diagnosis and potential legal abortion to prevent suffering. Risk factors include maternal dietary choices and genetic mutations, particularly in the Sonic Hedgehog (SHH) gene. Awareness and education regarding these risks are crucial for prospective parents, as early detection through ultrasound can guide management strategies and reduce harm to both the newborn and the mother.

1. Introduction

Cyclopia is an extremely rare congenital disorder that results from a genetic abnormality during early fetal development. It occurs in approximately 1 in every 13,000 to 20,000 live births, though many cases go undetected as they often lead to miscarriage. The primary characteristic of cyclopia is the failure of the eye orbits to properly divide into two separate cavities, causing the eyes to either merge into a single structure or be positioned closely together. This abnormality is typically part of a larger condition known as holoprosencephaly, where the brain fails to divide into two distinct hemispheres.[1,2] Cyclopia affects both humans and animals, including species such as sheep, cattle, and cats. In addition to eye malformations, those with this condition usually exhibit severe facial deformities. Most commonly, the nose is underdeveloped or absent, sometimes appearing as a proboscis located above the eye region. The outer ears may be malformed, with structures that are bent, folded, or collapsed. A polyp, typically ranging from 3 to 5 cm, often forms at the center of the brain, contributing to additional neurological complications. Because cyclopia is usually associated with profound defects in brain development, most affected embryos do not survive to term. If they are born, the chances of survival are extremely low, and infants typically die shortly after birth due to respiratory failure and other severe complications.[3]

2. Case Report

A 31-year-old woman, G2P1A0, came to Community Health Center Lingga, Sungai Ambawang Sub-district with a gestational age of 27 weeks with complaints of uterine contraction and bleeding from the birth canal since 1 day

* *Corresponding author*.
E-mail address: hee.tooth@gmail.com (N. Ambra).

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before admission. Previously, the patient had been to an obstetrician and gynecologist for an ultrasound at 22 weeks of gestation and was said to have an abnormality in the fetal head. A single fetus was found in cephalic presentation with an estimated fetal weight of 500 grams and a fetal heart rate of 158 times per minute. From the image of the fetal head, a single-lobed brain, facial abnormalities, and suspected cyclopia were found. The patient had never been sick during this pregnancy; there was no history of birth defects in the family. The patient had been advised to consult an obstetrician and gynecologist but did not do so. A vaginal touch examination was performed; the cervix was 5 cm open, the head dropped to Hodge degree 3, and uterine contractions were adequate. Based on the patient's condition, vaginal delivery was planned. 6 hours later, a baby boy was born with a birth weight of 800 grams, a birth length of 40 cm, and an APGAR score of 2/3. The fetus had only 1 eye with a trunk in the frontal area and was polydactyly. The baby was admitted to the perinatologi but died after 2 hours of treatment. The mother went home a day later in good health.

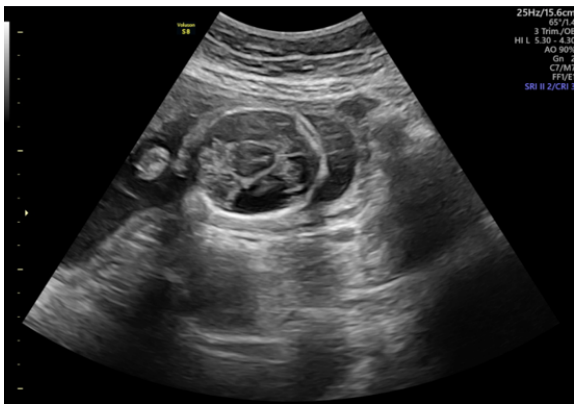

Figure 1: Ultrasound of the fetal head shows accumulation of fluid in the cranial cavity.

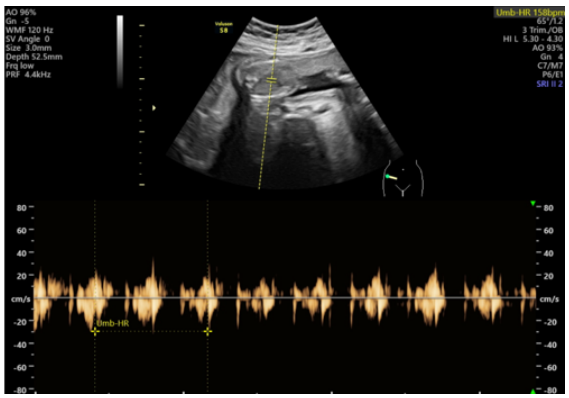

Figure 2: Ultrasound examinationfetal heart rate 158 beats per minute.

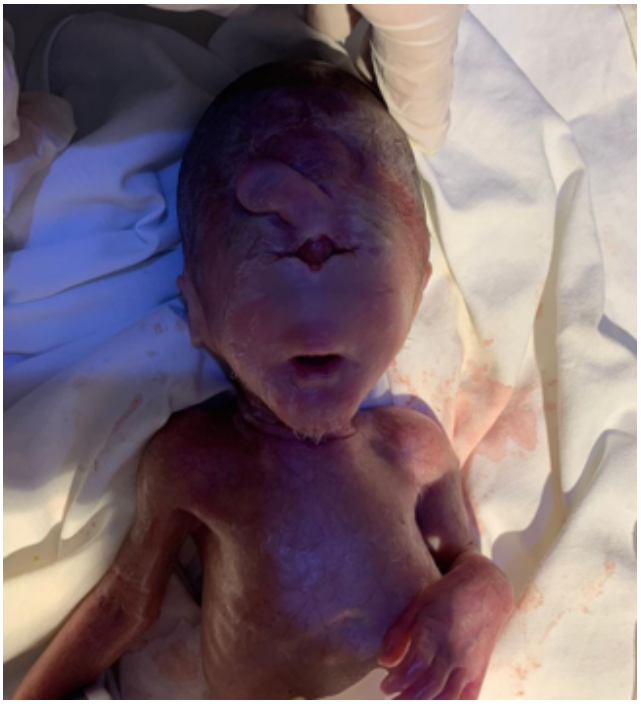

Figure 3: Thefetus after birth appears to have only one eye and has a proboscis in the frontal.

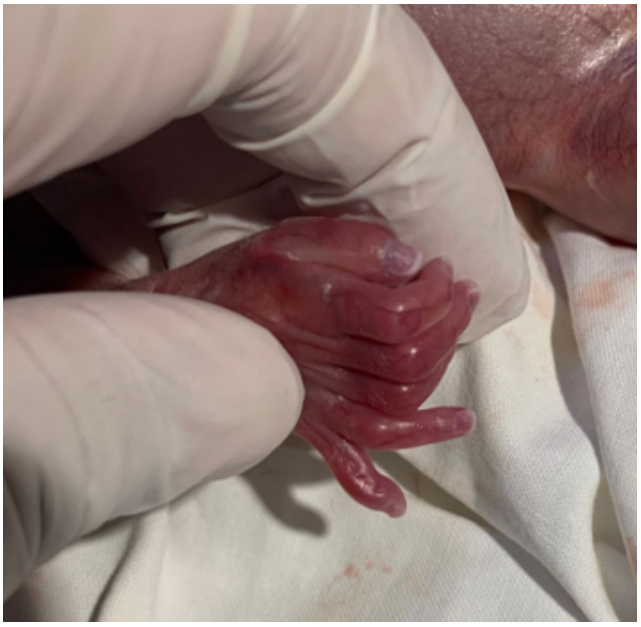

Figure 4: Polydactyly is seen on the fingers.

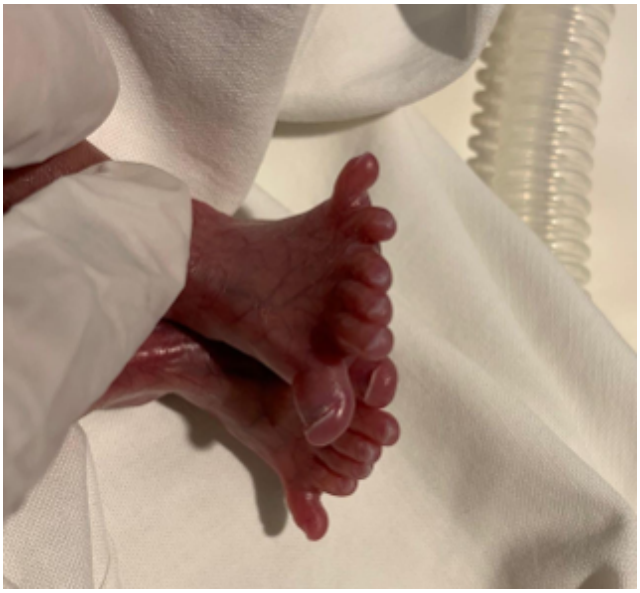

Figure 5: Polydactyly is seen on thetoes.

3. Discussion

Holoprosencephaly is a common brain malformation often associated with facial anomalies, such as closely spaced eyes, microcephaly, cleft lip, and cleft palate. This condition arises from the incomplete development and division of the prosencephalon (fetal forebrain) into left and right hemispheres, leading to a single-lobed brain structure and severe craniofacial deformities. In many cases, the severity of the abnormalities results in neonatal death prior to birth. Holoprosencephaly is categorized into three subtypes: alobar, semi-lobar, and lobar.[4] The alobar subtype features an undivided brain with significant facial defects, while the semi-lobar subtype exhibits partial division, resulting in a moderate form of the disorder. In lobar holoprosencephaly, the brain is divided into two hemispheres, but minor structural abnormalities are still present. Milder cases may present with craniofacial defects such as microcephaly, hypotelorism, a flat nasal bridge, and abnormal teeth.[5] Cleft lip is the least severe facial anomaly, whereas cyclopia characterized by a single eye and an underdeveloped nose is the most severe. Cyclopia is hereditary, and families should be informed about the increased risks for relatives considering having children. This case report discusses a female fetus born at 37 weeks and 5 days to a 44-year-old mother. The newborn lacked a nose and exhibited micrognathia but showed no signs of cleft lip or palate. The infant's skin appeared cyanotic, and a chest X-ray indicated complete collapse of the right lung, necessitating immediate chest tube insertion.[4–6]

Despite continuous ventilation, the newborn did not survive. An MRI revealed features of locoprosencephaly, including a malformed corpus callosum and an azygous anterior cerebral artery. Since there is no cure for holoprosencephaly, legal abortion may be considered to prevent further suffering for both the newborn and the mother. Identifying risk factors and educating parents are vital prevention strategies. Consumption of certain plants during pregnancy is linked to an increased risk of cyclopia and should be avoided. The Sonic Hedgehog (SHH) gene, located on chromosome 7, plays a critical role in brain development. Mutations in the SHH and PAX6 genes can lead to cyclopia.[5] Environmental and genetic factors such as multiple pregnancies, gestational diabetes, infections, and exposure to certain toxins can also increase the risk. Cyclopia can often be detected via ultrasound during pregnancy, usually between the third and fourth weeks. Early diagnosis and management are essential for minimizing harm to both the newborn and the mother.[6,7]

4. Conclusion

cyclopia represents a severe congenital disorder linked to holoprosencephaly, resulting in significant facial and neurological abnormalities. Early diagnosis through ultrasound is vital for managing affected pregnancies and informing parents of potential outcomes. Given the high mortality rate associated with this condition, discussions about legal abortion may be necessary to alleviate suffering for both the infant and the family. Understanding the genetic and environmental risk factors is essential for prevention, emphasizing the importance of prenatal care and education. Ultimately, awareness and timely intervention can help mitigate the impact of cyclopia and holoprosencephaly on affected families.

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6. Conflict of Interest

None.

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Author's biography

Nalmiades Ambra, Theo https://orcid.org/0009-0005-1516-5021

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