Case report

Cyclopia baby: Congenital lethal malformation: Rare case report

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ABSTRACT

Introduction and importance: Cyclopia is a rare congenital disorder characterized by facial abnormalities. It is the most severe form of alobar holoprosencephaly. This syndrome affects the embryos that are either aborted or stillborn upon delivery or, at best, die shortly after birth.

Case report: Patient aged 27 years, third gesture, third pare, admitted for premature delivery of 7 months. After labor management, she gave birth 4 h after admission to the maternity ward of a living newborn female weighing 1100 g. The newborn's Apgar score could not be calculated. In the initial physical examination, an eye and a 4-cm proboscis were seen in the middle of the forehead. The newborn had no nose, and his outer ears were normal.

Clinical discussion: Cyclopia is a rare deformity. It is considered to result from the fusion of two optic grooves, because of defective development of the ventral diencephalon (holoprosencephaly).

Conclusion: Prenatal diagnosis by ultrasound examination might help in the detection of cyclopia and in the prevention of complications associated with such a condition.

1. Introduction

Cyclopia is a rare and lethal complex human malformation. It is the most severe form of alobar holoprosencephaly, resulting from incomplete cleavage of prosencephalon into right and left hemispheres occurring between the 18th and the 28th day of gestation.

Approximately 1.05 in 100,000 births are identified as infants with cyclopia, including stillbirths [1].

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 [2]. Extra cranial malformations described in stillbirths with cyclopia include polydactyly, renal dysplasia, and an omphalocele.

This syndrome mostly affects the embryos that are either aborted or stillborn upon delivery or, at best, die shortly after birth [3]. We report this rare case to create awareness among people about the early diagnosis of fetal anomalies if present and more emphasis should be given to regular antenatal consultations.

This work has been reported with respect to the SCARE 2020 criteria [4].

2. Case report

Patient aged 27 years, third gesture third pare, mother of two children alive by vaginal route was admitted in the labor room of the Ibn Rochd university hospital center of Casablanca for a premature delivery on badly followed pregnancy presumed at 7 months. On questioning the parturient, there was no notion of consanguinity with her spouse. The parents did not mention the history of congenital anomalies or close family marriage in their relative. There was no notion of taking medication or plants during the pregnancy. The fasting blood sugar level was within normal limits during the pregnancy. No obstetrical ultrasound was performed during her pregnancy.

On admission, the patient was conscious, the blood pressure was 12/6 cmHg, the urine dipstick did not show any proteinuria. Her cervix was dilated to 4 cm with intact membranes.

On obstetrical ultrasound, we found a progressive monofetal pregnancy, with positive cardiac activity, cephalic presentation, microcephaly with a BIP corresponding to 24 weeks, i.e. lower than the third percentile (Fig. 1).

There was an associated hydramnios with a large cistern that was 13 cm, the placenta was fundal.

After labor management, she gave birth 4 h after admission to the maternity ward of a living newborn female weighing 1100 g. The newborn's Apgar score could not be calculated.

In the initial physical examination, an eye and a 4-cm proboscis were seen in the middle of the forehead. The newborn had no nose, and his
outer ears were normal (Figs. 2 and 3).

A cord sample was taken, the parents refused to do a karyotype.

The newborn expired within 06 h after birth. Unfortunately, her parents did not give consent for an autopsy.

3. Discussion

This case report we presented is a unique case with severe form of holoprosencephaly with cyclopia diagnosed in the postnatal period in a neonate born in Ibn Rochd university hospital center of Casablanca in Morocco.

Cyclopia is a rare deformity. It is considered to result from the fusion of two optic grooves, because of defective development of the ventral diencephalon (holoprosencephaly) [5].

Cyclopia is the severest facial expression of the holoprosencephaly syndrome [6]. Typically, the nose is either missing or replaced with a non-functioning nose in the form of a proboscis. Such a proboscis generally appears above the central eye, or on the back, and is characteristic of a form of cyclopia called rhinencephaly or rhinocephaly.

Parents of neonates with cyclopia should warn their first-degree relatives who may be forming a family about the increased risk of cyclopia or other milder forms of holoprosencephaly [7].

Cyclopia can be diagnosed using ultrasonography while the fetus is growing inside the uterus. [8]. This congenital disorder occurs between the third and fourth weeks of pregnancy, and conducting medical ultrasonography after this time can usually show obvious signs of cyclopia or other forms of holoprosencephaly.

The exact cause of this rare malformation is left unknown but research revealed there are multiple risk factors including teratogenic exposures and genetic causes. Among the possible teratogenic exposures which shown increased risk factor in animal model includes long use of

4. Conclusion

Prenatal diagnosis by ultrasound examination might help in the detection of cyclopia and in the prevention of complications associated with such a condition. However, in developing countries where women do not receive regular antenatal care and do not undergo prenatal diagnosis, such cases will go undetected.

Provenance and peer review

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Consent

Written informed consent for publication of their clinical details and/or clinical images was obtained from the parents of the newborn.

Ethical approval

I declare on my honor that the ethical approval has been exempted by my establishment.

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Declaration of competing interest

The authors declare having no conflicts of interest for this article.

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