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Case Report

Cyclopia Discovered in First Trimester Ultrasound

Sirine Abdelkafi, Rahma Ben Msarra, Dora Trabelsi, Hana Hakim*, Khaled Trigui and Kais Chaabane

Department of Gynecology and Obstetrics, University of Medicine of Sfax, Hedi Chaker Hospital, 3029, Sfax, Tunisia

***Corresponding Author:** Hana Hakim, Department of Gynecology and Obstetrics, University of Medicine of Sfax, Hedi Chaker Hospital, 3029, Sfax, Tunisia.

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Abstract

Cyclopia is a rare malformation of the face. We report the case of cyclopia diagnosed on the first trimester ultrasound performed at 12 weeks of gestation. Chromosome analysis revealed Trisomy 13 (47, XY,+13). Therapeutic abortion was indicated and accepted by the patient.

Keywords: Pregnancy; Ultrasound; Cyclopia; Fetal; Malformation

Introduction

In Greek mythology, one-eyed creatures belonged to a primordial race of giants called cyclocephalus. In the scientific world, cyclopia is a non-viable rare genetic anomaly. The incidence of this pathology is approximately 1,05 per 100.000 births [1]. Major malformations have an important psychological impact on the parents [2], which is why a meticulous morphological examination is necessary during the first trimester ultrasound to detect chromosomic anomalies and malformations early on [3].

Patient and observation

Cyclopia is a rare malformation of the face [4]. It represents the most severe form of holoprosencephaly. It is caused by the lack of development of the frontal bud, which is a unique medial bud raised by the cephalic extremity of the neural tube, while the rostral neuropore is closing. This pathology is part of the ectroprosopia anomalies [4]. Three types of ocular malformations were observed within the scope of cyclopia: one eye (monophthalmia), two fused ocular globes (synophthalmia) or the absence of both ocular globes (anophthalmia) [5].

What's original in our case is that we diagnosed this pathology before the delivery, at the time of the first trimester ultrasound. Therapeutic abortion was decided. We report the case of a 30-year-old patient without notable medical history, whose blood type was O positive. She was gravida 2, para 1 (G2P1), her past obstetrical history included one prior pregnancy with the delivery of a healthy baby by cesarean section for non-reassuring fetal heart rate pattern. There was no consanguinity between the patient and her husband, and no history of malformations in the family.

The first trimester ultrasound, performed at 12 weeks of gestation showed one ocular globe, monophthalmia, a supraorbital proboscis, an alobar holoprosencephaly (Figure 1), a major hydrocephalus, and an absent nasal bone (Figure 2).

The rest of the morphological examination showed a bilateral hand polydactyly. Nuchal translucency was 3.9 mm, above the 95th centile (Figure 2). A trophoblast biopsy was performed, and the fetal karyotype was (47,XY,+13), confirming Trisomy 13 diagnosis. Therapeutic abortion was accepted by the patient and her husband (Figure 3).

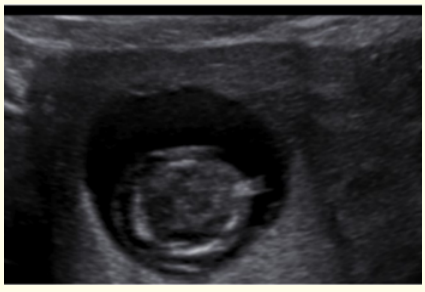

Figure 1: Alobar holoprosencephaly and proboscis.

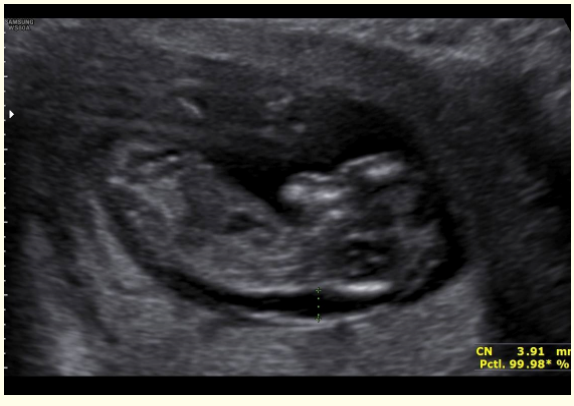

Figure 2: Nuchal translucency, absent nasal bone.

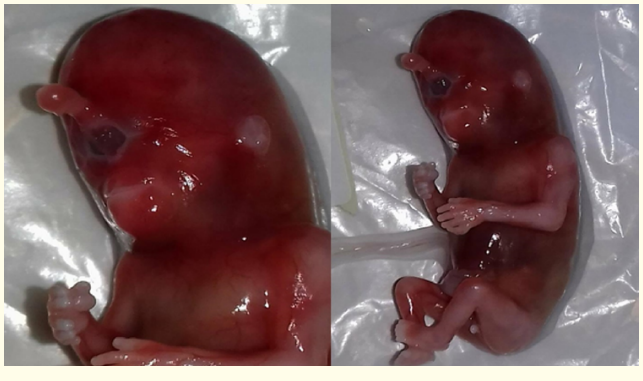

Figure 3: Male fetus presenting cyclopia, absent nasal bone, proboscis, and polydactyly.

Conclusion

Cyclopia is the most severe form of craniofacial anomalies associated with alobar holoprosencephaly. Early prenatal diagnosis is possible via the first trimester ultrasound. The signs we should look for are monophthalmia, absent nasal bone, supraorbital proboscis and alobar brain development. The common association between holoprosencephaly and chromosomal aneuploidies (Trisomy 13, Trisomy 18) warrants systematic fetal karyotyping.

Conflict of Interest

The authors declare no conflict of interest.

Bibliography

1. Salama GS., *et al*. "Cyclopia: a rare condition with unusual presentation - a case report". *Clinical Medicine Insights: Pediatrics* 9 (2015): 19-23.
2. Rajon A-M., *et al*. "Répercussions du diagnostic périnatal de malformation sur l'enfant et ses parents : approche métapsychologique à partir de l'étude longitudinale de 30 familles". *Psychiatrie de l'enfant* 49.2 (2006): 349.
3. Chelli D., *et al*. "ECHOGRAPHIE DU PREMIER TRIMESTRE : UN OUTIL DE DÉPISTAGE PRÉCOCE DES MALFORMATIONS FŒTALES ET DES ANOMALIES CHROMOSOMIQUES". *Tunis Media* 87 (2009): 7.
4. Amadou D and Youssouf K. "La cyclopie, maformation rare du visage dans un centre de santé de référence de Bamako à propos d'un cas". *Pan African Medical Journal* 33 (2019).
5. Amadou DA., *et al*. "Holoprosencephalie alobaire dans un contexte de syndrome polymalformatif: apport de l'imagerie, à propos d'un cas". *Pan African Medical Journal* 15 (2013).