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Case Report

Cyclopia Associated with Microcephaly, Hydrocephaly and Clubfoot: A Case Report from Burkina Faso, West Africa

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Abstract

Cyclopia is the most severe form of alobar holoprosencephaly, characterized by the fusion of the two orbits into one. It is a genetic disease incompatible with life. It is most often associated with other craniofacial malformations such as microcephaly, cleft lip and palate, nasal agenesis and facial dysmorphia. The incidence of cyclopia varies from 1/13,000 to 1/20,000 births, and the etiology of this malformation has yet to be elucidated. These include genetic factors, multiparity, female gender, unexplained miscarriage, gestational diabetes, infections during pregnancy, UV radiation, smoking and alcohol consumption. It is probably less diagnosed in developing countries, where pregnancies are not normally monitored, and antenatal ultrasound is less common. We report the case of a 22-year-old pregnant woman, third gesture and two pares with two live children. She came on her own for a third-trimester ultrasound. She had a spontaneous pregnancy. She reported no previous history of a spontaneous pregnancy. Morphological ultrasound revealed cyclopia associated with microcephaly and hydrocephaly. We obtained informed consent from the couple. The patient exploded with a female fetus weighing 700 g, her cranial perimeter at 18 cm and thoracic perimeter at 34 cm. A clubfoot was noted on expulsion of the fetus, which is a particular feature of our case.

Keywords

Cyclopia, Holoprosencephaly, Hydrocephaly, Burkina Faso

1. Introduction

Cyclopia is an extremely rare genetic facial malformation characterized by the fusion of the orbits and the presence, more often than not, of a single eye in the middle of the forehead. It is the most severe form of holoprosencephaly, most often associated with other craniofacial malformations [1]. The incidence of cyclopia varies from 1/13,000 to

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1/20,000 births [2]. Holoprosencephaly is a disorder in the development and closure of the prosencephalon into left and right hemispheres [3]. The etiology of this malformation remains unclear, but certain factors may increase the risk of its occurrence. These include genetic factors, multiparity, female gender, unexplained miscarriage, gestational diabetes, infections during pregnancy, Ultraviolet radiation (UV radiation), smoking and alcohol consumption [4]. This type of alobar holoprosencephaly is incompatible with life [5]. In countries with limited resources, diagnosis is most often postnatal, as pregnancies are insufficiently monitored with ultrasound scans, which are not accessible to all pregnant women [1, 6]. We present here a case of antenatal cyclopia associated with nasal agenesis, hydrocephalus and left clubfoot.

2. Case Presentation

This is a 22-year-old housewife living in Nobili in the south-central region of Burkina Faso. She is fourth gesture, third pare with three living children who are in apparent good health. She is married to a farmer who allegedly handles pesticides of a nature unspecified by the patient. There is no known history of malformations in the household, and she does not consume cola, tobacco or alcohol. The pregnancy occurred spontaneously. No folic acid was taken prenatally or during the first trimester. She is seen for a third-trimester ultrasound as part of her prenatal follow-up. She had received an ultrasound report for the second trimester, which she had not honored due to lack of funds.

The ultrasound performed on May 14, 2024 showed a discrepancy between the estimated gestational age based on cephalic measurements and other fetal biometry measurements. Cephalic perimeter. The biparietal diameter was 51 mm or 21 weeks of amenorrhea, the cephalic circumference 194 mm or 21 weeks of amenorrhea. The femoral diameter was 53 mm, i.e. 27 weeks' gestation, and the abdominal circumference was 229 mm, i.e. 28 weeks' gestation. Morphological analysis revealed dilation of all ventricular cavities suggestive of hydrocephalus (figure 1), nasal agenesis and a single orbit containing an eyeball (figure 2). The fetus was female. The rest of the morphological examination was unremarkable.

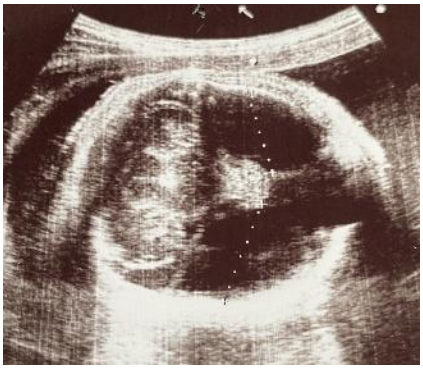
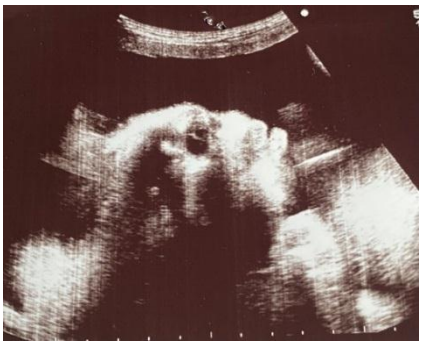


Figure 1. *Image showing hydrocephalus.*



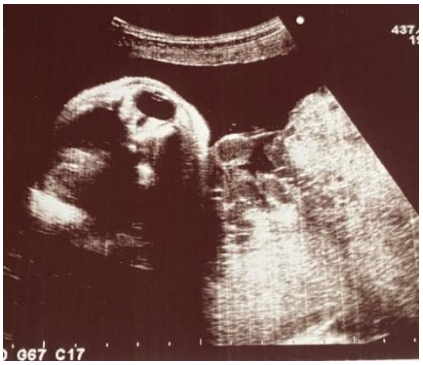


Figure 2. *Image showing single orbit and eyeball.*

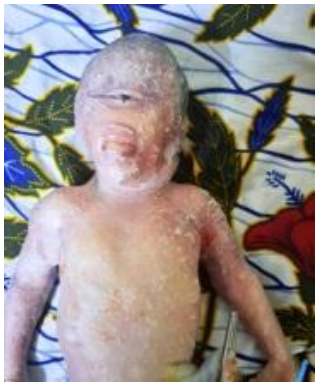


Figure 3. *Image showing cyclopia at fetal expulsion.*

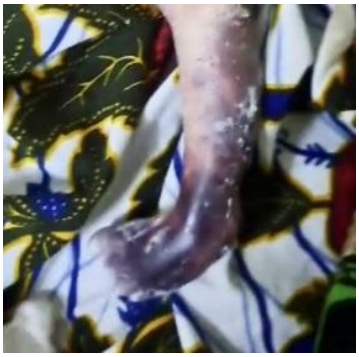


Figure 4. *Image showing clubfoot.*

The patient's management consisted of a medical termination of pregnancy after obtaining informed consent from the parents.

At expulsion, in addition to the cyclopia (figure 3), the fetus presented with a left clubfoot (figure 4). The fetus weighed 700 g, had a head circumference of 18 cm and a chest circumference of 34 cm.

3. Discussion

Holoprosencephaly comprises three subtypes of decreasing severity. In its alobar form, the brain is not divided in two and leads to severe craniofacial malformations such as cyclopia, the semi-lobar form where the brain is partially divided in two hemispheres leading to the moderate form of malformations and the alobar form in which the brain is divided in two hemispheres with minor defects of the disease. In the latter two forms, microcephaly, orbital hypotelorism, flat nasal bridge and abnormal anterior teeth may be present. Cleft lip represents the mildest form of these holoprosencephaly anomalies [7, 8].

Cyclopia is incompatible with life. In contrast, Meeker [9] described the case of a 5-year-old child living with cyclopia. However, this was a combination of cyclopia, lateral proboscis and a normal hemi-face.

Holoprosencephaly is most often associated with other malformations such as microcephaly, cleft lip and palate, nasal agenesis and facial dysmorphia [4, 10]. In Singh [11], cyclopia was associated with agnatia, synotia, astomia. In Carles [12], cyclopia was associated with otocephaly and other cerebral anomalies. In our case, cyclopia was associated with microcephaly, hydrocephaly and left clubfoot. In the literature, our case is the only one in which a clubfoot has been found in association with cyclopia.

In developing countries, pregnancies are often poorly or not at all monitored, and cyclopia is discovered in the delivery room when antenatal ultrasound could have described other malformations [1, 6].

4. Conclusion

The management of cyclopia, although rare, will require the popularization of early antenatal ultrasound in parturients. Regular follow-up of subsequent pregnancies is essential. Genetic counselling will help prevent further cases in the family.

Abbreviations

UV radiation	Ultraviolet Radiation

Author Contributions

Lompo Yemboado Dieudonne: Data curation, Writing – original draft
Kiemtore Sibraogo: Methodology, Writing – original draft
Traore Solo: Writing – review & editing
Sawadogo Zakaria: Project administration, Resources
Ouedraogo Mahamoudou: Visualization
Dao Blami: Writing – review & editing

Conflicts of Interest

The authors declare no conflicts of interest.

References

[1] La cyclopie: malformation rare du visage dans un centre de santé de référence de Bamako à propos d'un cas [Cyclopia : a rare facial malformation in a referral health center in Bamako : a case report]- The Pan African Medical Journal; Kampala Vol. 33, (2019). https://doi.org/10.11604/pamj.2019.33.261.19240

[2] Yanoff M, Sassani JW. Ocular Pathology-E-Book [Internet]. Elsevier Health Sciences; 2023 [cité 19 mai 2024]. Disponible sur: https://books.google.com/books?hl=fr&lr=&id=1bPxEAAAQBAJ&oi=fnd&pg=PP1&dq=M.+Yanoff,+J.W.+Sassani.+Ocular+Pathology-E-Book,+Elsevier+Health+Sciences,+2018.&ots=nruoUxhgaE&sig=aduflEbBO40I2Ih4oPjqdxlOVHg

[3] Kruszka P, Muenke M. Syndromes associated with holoprosencephaly. American Journal of Medical Genetics Part C: Seminars in Medical Genetics. Am J Med Genet c Semin Med Genet. 2018 Jun; 178(2): 229-237. https://doi.org/10.1002/ajmg.316202

[4] Parizad N, Faraji N, Hassanpour A, Goli R, Rostami S, Amanollahzadeh A. Cyclopia, a newborn with a single eye, a rare but lethal congenital anomaly: A case report. International Journal of Surgery Case Reports. Int J Surg Case Rep. 2021 Nov; 88: 106548. Published online 2021 Nov 4. https://doi.org/10.1016/j.ijscr.2021.106548

[5] Sarma V. OCULAR ABNORMALITIES OF THE FOETUS WITH SPECIAL REFERENCE TO CYCLOPIA*. Br J Ophthalmol. Br J Ophthalmol. 1963 Apr; 47(4): 193–202. https://doi.org/10.1136/bjo.47.4.193

[6] Diaouga HS, Yacouba MC, Garba RM, Idi N, Nayama M. La Cyclopie (Nouveau-N é avec un Seul ØEil): À Propos d'un Cas au Niger. [Cyclopia (Newborn with only one eye) About a case in Niger] [cité 17 mai 2024]; Disponible sur: https://www.academia.edu/download/109575963/HSD_cyclopie.pdf

[7] Dubourg C, Kim A, Watrin E, de Tayrac M, Odent S, David V, et al. Recent advances in understanding inheritance of holoprosencephaly. American Journal of Medical Genetics Part C: Seminars in Medical Genetics. https://doi.org/10.1002/ajmg.c.31619

[8] Roessler E, Hu P, Muenke M. Holoprosencephaly in the genomics era. American Journal of Medical Genetics Part C: Seminars in Medical Genetics. https://doi.org/10.1002/ajmg.c.31615

[9] MEEKER LH, AEBLI R. CYCLOPEAN EYE AND LATERAL PROBOSCIS WITH NORMAL ONE-HALF FACE: Report of a Case. Archives of Ophthalmology. 1 août 1947; 38(2): 159‑73.

[10] Mansouri Hattab N, Lahmiti S, Bouaichi A, Hiroual A, El Bouihi M, Fikry T. Les fentes labio-palatines médianes: un diagnostic qui en cache un autre. [Median cleft lip and palate: one diagnosis hides another] Archives de Pediatrie. https://doi.org/10.1016/j.arcped.2010.11.007

[11] Singh K, Sharma S, Agarwal K, Kalra A. Cyclopia-Otocephaly-Agnathia-Synotia-Astomia Complex: A Case Report. Journal of Clinical Neonatology. https://doi.org/10.4103/jcn.JCN_23_18

[12] Carles D, Serville F, Mainguené M, Dubecq JP. Cyclopia-otocephaly association: a new case of the most severe variant of agnathia-holoprosencephaly complex. J Craniofac Genet Dev Biol. 1 janv 1987; 7(2): 107‑13.