

Case report of cyclopia baby - A rare lethal anomaly

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ABSTRACT

Cyclopia is a rare and lethal human malformation, with a single eye in the center of the forehead. We report the case of a woman aged 23 years (second gravida) referred to us at 30 weeks of pregnancy and delivered a stillborn weighing 1.02 kg. The baby had a single central eye, low set ears, omphalocele, and single umbilical artery. With advances in fetal imaging and availability of high-resolution ultrasound machines, it is surprising that such lethal malformations are still presenting after the period of viability. To reduce such incidences, more awareness and emphasis must be made out on early ultrasound diagnosis of fetal anomalies and regular antenatal visits.

Key words: *Cyclopia, Holoprosencephaly, Single eye*

Cyclopia (alobar holoprosencephaly [HPE]) is a rare and lethal human malformation. The term “cyclopia” comes from the word Cyclops, the single-eyed giants of Greek mythology. It is a serious median facio-cerebral developmental deformity in which the baby is stillborn or dies soon after birth [1]. Approximately 1.05 in 100,000 births, including stillbirths, are identified as cyclopean [2]. The prevalence of HPE is about 1 in 11,000–20,000 live births and 1 in 250 embryogenesis [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 visits.

CASE REPORT

A 23-year-old woman G2P1 (with one healthy child delivered at term by cesarean section) came to the department in her 30th week of pregnancy with good dates. She was referred for the problem of polyhydramnios, which was detected during ultrasonography. The patient had a history of second degree consanguinity. She was not a smoker or habituated to alcohol intake and had not taken any teratogenic drugs. There was no history of any infectious disease during pregnancy.

First ultrasound examination, which was done at 30 weeks showed microcephaly, single orbit, absent stomach bubble, omphalocele, single umbilical artery, and polyhydramnios. The couple was counseled for the grave prognosis. After 1 week, a patient came back with labor pain and delivered a stillborn male baby weighing 1.02 kg with multiple congenital anomalies. The following abnormalities were noted (Fig. 1): Single central eye, low set ear, omphalocele, and single umbilical artery.

DISCUSSION

HPE is a group of disorders arising from the failure of normal forebrain development during embryonic life. There are three forms of HPE: Alobar, Semilobar, and lobar varieties.

Alobar HPE is the most severe form, no division takes place in prosencephalon. The two lateral ventricles appear as a single ventricle. Thalamic fusion is observed. No interhemispheric fissure, optic tracts, or olfactory processes were seen. Corpus callosum does not exist. Facial development may also be affected as there may be cyclopia, proboscis, ethmocephaly, cebocephaly, and median cleft lip, and palate.

Semilobar HPE is an intermediate form. In this, there is partial segmentation of ventricles and partial fusion of thalami. Olfactory processes and corpus callosum are usually absent.

Lobar HPE is the mildest form in which the hemispheres are well split. However, there is a fusion in the rostral portion. Lateral ventricles are interconnected, albeit dilated. Corpus callosum is either normal or hypoplastic. In 80% of the cases, there are midline defects while 20% of the affected cases have a normal appearing face.

Cyclopia results from incomplete cleavage of prosencephalon into right and left hemispheres, as a result, there is only one eye. That eye is centrally placed in the area normally occupied by the root of the nose. 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 [4,5].

There are three types of eye deformities seen in cyclopia: One eye (monophthalmia), two fused eyeballs (synophthalmia),

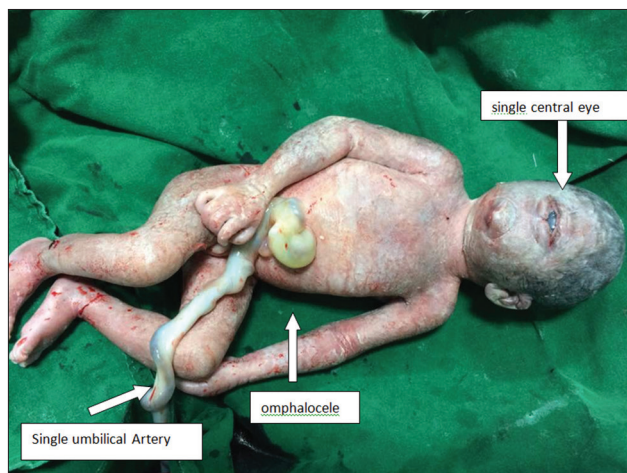


Figure 1: Cyclopia baby

or complete absence of eyeballs (anophthalmia). The fetus in this case report had monophthalmia (one eye). Extracranial malformations described in stillbirths with cyclopia include polydactyl, renal dysplasia, and an omphalocele [6]. The majority of cases with additional anomalies are diagnosed to have chromosomal abnormalities.

The etiology of HPE includes genetic and environmental factors. Among the environmental causes, there are maternal diabetes mellitus, maternal alcoholism, *in utero* infections with Cytomegalovirus, rubella or toxoplasma, and some drugs (retinoic acid and cholesterol synthesis inhibitors) [7]. HPE can be transmitted in an autosomal dominant way. Mutation of SHH gene is the most frequent cause of familial HPE [8]. Furthermore, HPE is associated in 40% of cases with numerical chromosomal anomalies, the most frequent one being trisomy 13 [8]. HPE can also be associated in about 25% of the cases with several defined multiple malformation syndromes with a normal karyotype such as Smith-Lemli-Opitz, Pallister Hall, or velo-cardio-facial syndrome [7]. However, in the presented case, no risk factors could be identified.

CONCLUSION

Advances in fetal imaging and availability of high-resolution ultrasound machines have made it possible to diagnose various lethal structural defects like cyclopia in first trimester itself. In spite of all these advancements, such anomalies are being detected late in the second and third trimester because of irregular investigations and follow-up to the doctor. Hence, awareness should be created among people for early diagnosis of fetal anomalies, if present and more emphasis should be given for regular antenatal visits.

REFERENCES

1. Arathi N, Mahadevan A, Santosh V, Yasha TC, Shankar SK. Holoprosencephaly with cyclopia-Report of a pathological study. *Neurol India* 2003;51:279-82.
2. Cannistra C, Barbet P, Parisi P, Iannetti G. Cyclopia: Radiological and anatomical craniofacial postmortem study. *J Craniomaxillofac Surg* 2001;29:152.
3. Moog U, De Die-Smulders CE, Schrandner-Stumpel CT, Engelen JJ, Hamers AJ, Frints S, *et al.* Holoprosencephaly: The maastricht experience. *Genet Couns* 2001;12:287-98.
4. DeMyer W, Zeman W. Alobar holoprosencephaly (arhinencephaly) with median cleft lip and palate: Clinical, electroencephalographic and nosologic considerations. *Confin Neurol* 1963;23:1-36.
5. Graham D. Rhinencephaly. In *Online Medical Dictionary*; 2007. Available from: <https://www.elsevier.com/books/dorlands-illustrated-medical-dictionary/dorland/978-1-4160-6257-8>. [Last retrieved on 2008 Jul 23].
6. Salama GS, Kaabneh MA, Al-Raqad MK, Al-Abdallah IM, Shakkoury AG, Halaseh RA. Cyclopia: A rare condition with unusual presentation-a case report. *Clin Med Insights Pediatr* 2015;9:19-23.
7. Dubourg C, Bendavid C, Pasquier L, Henry C, Odent S, David V. Holoprosencephaly. *Orphanet J Rare Dis* 2007;2:8.
8. Poenaru MO, Vilcea ID, Marin A. Holoprosencephaly: Two case reports. *Medica (Buchar)* 2012;7:58-62.

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