

A Case of Cyclopia

Fahri YILMAZ, Ali Kemal UZUNLAR, Nihal KILINÇ, Gökhan BAYHAN, Fuat GÜRKAN, Murat YAYLA
Dicle Üniversitesi Tıp Fakültesi Patoloji ve Kadın Hastalıkları ve Doğum Anabilim Dalı-DİTARBAKIR

ÖZET

BİR SIKLOPI OLGUSU

Gerçek siklopi organogenetik gelişim esnasında ortaya çıkan nadir bir anomali olup iki ayrı gözün gelişiminin baskılanmasıdır. Biz siklopi ile birlikte diğer anomalileri olan bir fetusu rapor ettik. Ayrıca siklopinin olası oluşum mekanizması ilgili literatürle birlikte tartışıldı.

Anahtar Kelimeler: Siklopi, fetus, holoprosensefali.

SUMMARY

A CASE OF CYCLOPIA

True cyclopia is a rare anomaly in which the organogenetic development of the two separate eyes is suppressed. We reported a fetus with an association of cyclopia with other anomalies. The possible mechanism of the histogenesis is discussed, together with a review of the relevant literature.

Key Words: Cyclopia, fetus, holoprosencephaly.

Cyclopia is an unusual anomaly in which the anterior brain and the midline mesodermal structures develop anomalously. The orbital region is grossly deformed, resulting in the formation of a central cavity eipseudoorbitii, with absence of nasal cavity and presence of a rudimentary proboscis above the pseudo-orbit. If two globes are found in a different degree of fusion in the pseudo-orbit, the condition is called synophthalmos. A much rarer anomaly is true cyclopia, wherein only eye is present (1,2).

CASE REPORT

This infant was born to a 23-year-old G3P2 woman and her 25-year-old husband. The marriage was not consanguineous. The first two children were normal. All investigations of the mother were normal. At third trimester of pregnancy, ultrasound investigation confirmed the suspected polyhydramnios and at the same time documented marked hydrocephaly and holoprosencephaly of the fetus (Fig. 1). Amniocentesis was performed, and 500 ml of amniotic fluid was withdrawn to reduce the polyhydramnios. The fetus was delivered at 33 weeks by spontaneously. After delivery, the cyclopic infant showed severe apnea and bradycardia, the child died after 5 minutes. His length was 40 cm, weight was 1400 g, and head circumference was 30 cm. There was a midline frontal proboscis, single midline orbit with single eye of fused eyes, a chin-like structure at the lower part of the face (Fig. 2). Aside from having anal atresia and hypoplastic male genitalia, there was meningomyelocel. Other anomalies

included short right femur and tibia, right club-foot and bilateral pes equino varus (Fig. 3).

At autopsy, there was cyclopia and a proboscislike mass above the eye. The proboscis measuring 15 mm length and 10 mm diameter with a single blind end orifice was attached to the superior aspect of the orbit. The nose was absent. There was a single median ventricular cavity and (alobar) holoprosencephaly, but no olfactory apparatus no optic chiasma, and no optic tracts or pituitary gland. Only are median connection was seen between a single optic foramen and the eyeball, which was presumed to be the sheaths of the optic nerve.

The other viscera were macroscopically normal. Microscopically, was no a pathologic finding. The placenta and umbilical cord were macroscopically and histologically normal. Chromosomal studies taken from the cordocentesis showed a normal karyotype (46-XY).

DISCUSSION

Cyclopia is a rare deformity. It is considered to results from the fusion of two optic grooves, because of defective development of the ventral diencephalon (holoprosencephaly). Cyclopia is commonly divided into true cyclopia and synophthalmia (2). True cyclopia is very rare, and the two eyes are completely fused (3). Synophthalmia is more commonly seen and is characterized by fusion of two eyes by varying degrees (4). We evaluated our case as a true cyclopia.

Defective genetic and environmental factors have been associated with cyclopia (5,6). Agents that have been shown to induce cyclopia in animals include magnesium salt, alcohol, lithium chloride, retinol, and

Corresponding Address: Dr. Fahri Yılmaz

Dicle Üniversitesi Tıp Fakültesi, Patoloji ABD - DİTARBAKIR

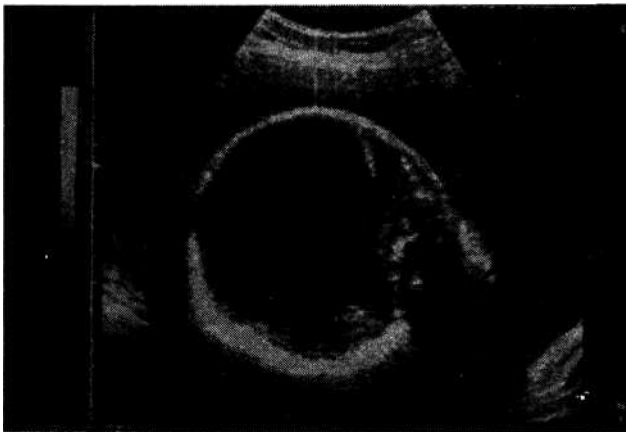


Fig 1: The sonographic picture of the skull of the 33-week-old fetus.

radiation (7). In human beings, environmental factors associated with this deformity include ionizing radiation, contraceptives, viremia, salicylate, rubella vaccine, antibiotics, and aminopyrine (4,8).

Warkany and Takacs (9) administered high doses of salicylates to pregnant rats and obtained a variety of anomalies, particularly of the nervous system, including craniorachioschisis. All report in man remains short of convincing in the absence of prospective studies (10). A lucid discussion of the problem may be found in a paper by Benawra et al (11). They presented a case of cyclopia with multiple other anomalies born to a mother who took 3 to 4.5 g of aspirin daily during the first trimester.

Genetic errors with chromosomal abnormalities such as trisomy-D, monosomy-G mosaicism, translocation affecting chromosome 3 and group C chromosome, and chromosome 10 short arm deletion have also been recorded in cyclopia (1,12). Taysi and Tinaztepe (13) suggested a classification of two main aetiological groups. In the first group, there were only cephalic malformations and the karyotype was nor-



Fig 2: The fetus with cyclopia having midline frontal proboscis above the eyeball.



Fig 3: The fetus showing meningocele, short right femur and tibia, right clubfoot and bilateral pes equinovarus.

mal. In the second group there were cephalic as well as extracranial malformations with anomalous karyotype, mostly trisomy-D. We detected that the case had cephalic malformation including a single median ventricular cavity and (alobar) holoprosencephaly, and that there was no olfactory apparatus, no optic chiasma, and no optic tracts or pituitary gland. In addition, in our case, the extracranial defects having anal atresia, hypoplastic male genitalia, meningocele, short right femur and tibia, right clubfoot and bilateral pes equinovarus were detected. However, in our case the karyotype was normal.

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