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Case Report / Olgu Sunumu

A Term Fetus With Alobar Holoprosencephaly and Cyclopia: Case Report

Alobar Holoprosensefali ve Siklopili Term Bir Fetus: Vaka Sunumu

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Summary

Alobar holoprosencephaly(HPE) is a rare, severe, complex human brain malformation. We describe a case of cyclopia with alobar HPE identified at 40 weeks of gestation by 2-dimensional (2D) and 3D transabdominal ultrasound (US).

A 27-year-old woman, gravida5, para4, abortion0, was referred to our department at 40 weeks of gestation; she was experiencing labor pains and had a nonreactive non-stress test. The mother had received no regular antenatal care, and no sonography was performed during pregnancy. Both 2D and 3D US revealed alobar HPE and cyclopia. The baby was born by vaginal delivery shortly thereafter and then died after birth. On examination, the face had a single, large median eye and no nose.

Alobar HPE and cyclopia can be diagnosed by US early during pregnancy. Early diagnosis is important to allow for early termination of pregnancy and to minimize the physiological and psychological impact of such anomalies on the mother and family.

Key words: Alobar Holoprosencephaly, Cyclopia, Diagnosis, Ultrasonography

Özet

Alobar holoprosensefali (HPE), insan beyninin nadir görülen ağır bir malformasyonudur. 40. gebelik haftasında 2 boyutlu (2D) ve 3D transabdominal ultrason (US) ile tanımlanan alobar HPE ve siklopili olguyu sunduk.

27 yaşında, gravida 5, parite 4, abortus 0, kadın hasta Sakarya Üniversitesi Kadın Hastalıkları ve Doğum Anabilim Dalı'na 40. gebelik haftasında doğum sancısı ile başvurdu; non stres test non reaktif izlendi. Hasta doğum öncesi düzenli bakım almamış ve gebelik sırasında sonografi yapılmamıştı. Hem 2D hem de 3D US' de alobar HPE ve siklopi mevcuttu. Bebek spontan vajinal doğum ile doğdu ve doğumundan kısa süre sonra öldü. Muayenesinde yüzde kirpikleri olan tek, büyük bir medyan göz mevcuttu ve burun olmadığı tespit edildi.

Alobar HPE ve siklopi, gebelik sırasında erken yapılan US ile teşhis edilebilir. Erken teşhis gebeliğin erken sonlandırılmasını sağlayarak, annede ve ailede bu anomalilerin fizyolojik ve psikolojik etkisini en aza indirgediği için önemlidir.

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Anahtar kelimeler: Alobar Holoprosensefali, Siklopi, Tanı, Ultrasonografi

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Introduction

Holoprosencephaly (HPE) is a rare and complex human cerebral malformation affecting forebrain development, resulting in incomplete or no differentiation of the forebrain into left and right hemispheres during early embryonic life. This impaired development of the forebrain structures is associated with a wide spectrum of craniofacial dysmorphisms ranging from anophthalmia, cyclopia or proboscis in the most severe cases, to midline cleft lip, a simple hypotelorism or even a complete lack of facial anomalies in the less severe HPE forms. HPE occurs in 1/16 000 live births[1] and 1/250 conceptuses[2]. It was classified into the following 4 categories by Demyer et al. based on the severity of the disease and the pathological characteristics of the forebrain: alobar, semilobar, lobar and a middle interhemispheric fusion variant[3].

Alobar HPE is the most severe form of HPE and is characterized by a complete absence of the interhemispheric fissure and the corpus callosum with a single cerebral ventricle and facial dysmorphism that may include cyclopia, proboscis, ethmocephaly and cebocephaly[4].

Cyclopia is a rare congenital eye deformity defined by a single midline orbit that contains ocular structures that can be monophthalmic, synophthalmic, or anophthalmic[5]. It is a severe craniofacial abnormality and is typically associated with alobar HPE or rarely with semilobar HPE. İt has been reported to occur in approximately 1 in 100,000 births[6].

We describe the identification of cyclopia with alobar HPE and arhinia at unfollowed 40 weeks of gestation using 2- (2D) and 3-dimensional (3D) transabdominal ultrasound (US).

Case Report:

A 27-year-old woman, gravida 5, para 4, abortion 0, was referred to our Obstetrics and Gynecology department at 40 weeks of gestation (according to her last menstrual period) for sonography. She was experiencing labour pains and had a nonreactive non-stress test (NST). Hydrocephalus was suspected at her last visit to the local medical clinic. The first fetal US revealed polyhydramnios, microcephaly and alobar holoprocencephaly. The biometric measurements were as follows: a biparietal diameter of 79.6 mm (32 week), an abdominal circumference of 334.9 mm (37+3 week), and a femur length of 75.4 mm (38+4 week).

The evaluation of the fetal anatomy using 2D US (GE Voluson®730Pro USA, 1-5 mHz transabdominal probe) showed abnormal development of the brain with cyclopia and the absence of a nasal septum. A fused thalami was observed, and no mid-line echo could be identified (Figure 1). We evaluated the fetal face using 3D US with a transabdominal transducer (GE Voluson®730Pro USA, 2-8 mHz transabdominal probe) and were able to detect cyclopia at 40 weeks of gestation (Figure 2). These findings are characteristic of alobar holoprosencephaly.

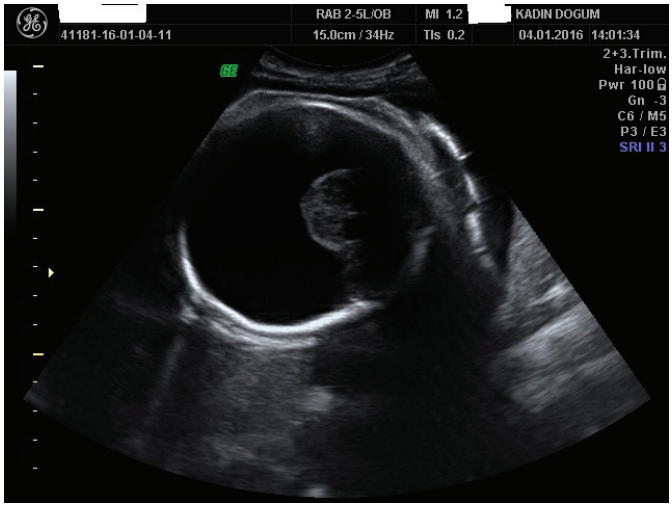

Figure 1: Shows the USG 2D Image (A fused thalami and no mid-line echo)

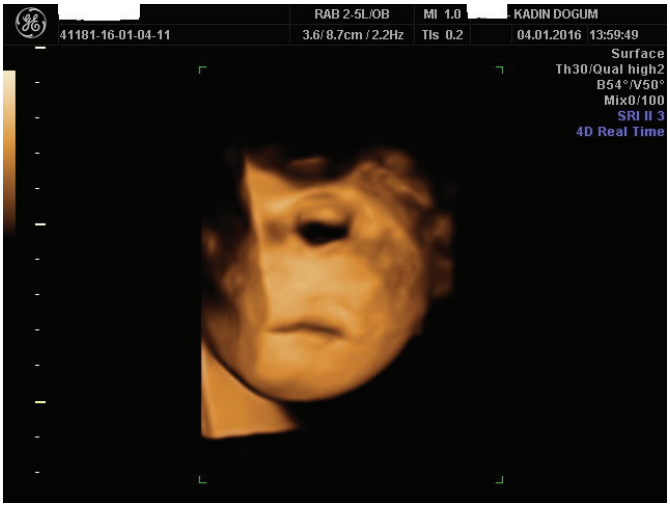

Figure 2: Shows the USG 3D Image (A 40-week fetus with cyclopia)

The mother had four uncomplicated pregnancies prior to this conception, all with normal deliveries. There was no history of exposure to chemicals, drugs, alcohol, smoking, radiation or other environmental hazards. The maternal history was unremarkable for any prenatal infections, trauma or any other chronic disease during pregnancy. There was no history of hereditary disease or chromosome disorders in either family. No significant obstetric or family history was elicited. The parents were 27 and 31, non-consanguineous and without dysmorphic features or congenital anomalies.

The mother had received no regular antenatal care, including US examination, throughout the pregnancy; hence, no diagnosis was made earlier.

A baby boy was subsequently born by normal, spontaneous vaginal delivery. He weighed 2600 g and had an APGAR score of 0 at 1 and 5 minutes after birth. The baby died soon after birth. The examination of the head revealed a large single centrally located eye with eyelash and micrognathia (Figure 3). The eyelids, eyebrows and nose were absent. Neither a cleft lip nor a cleft palate was noted. The ears had a deformity at a lower level. The appearance of the extremities were normal. A postmortem autopsy and chromosomal analysis were not performed as consent to these procedures was not given by the parents because of religious beliefs.

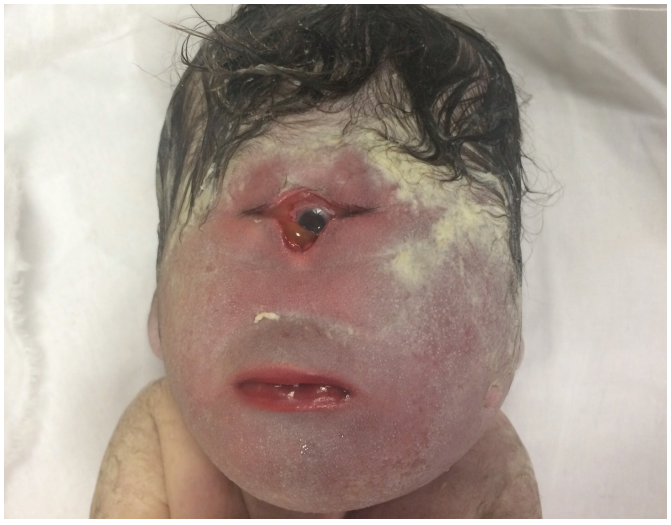

Figure 3: Fetal face with a large single centrally located eye and eyelash

Discussion:

The etiology of HPE is not clearly understood. Most cases are sporadic, but the etiology of this syndrome appears to involve a number of heterogeneous risk factors such as maternal diabetes mellitus, alcohol abuse, exposure to retinoic acid and cholesterol biosynthesis inhibitors, low maternal cholesterol levels, perinatal infections, chromosomal defects[7]. İn the present case, our patient had no history of exposure to environmental hazards and or prenatal infections. In addition, there was no consanguinity in the family.

Salama et al. reported a case with a live full-term newborn infant with cyclopia, including the typical facial manifestations as well as three extrafacial malformations, namely a huge omphalocele, bladder exstrophy, and ambiguous genitalia[8]. Ozden et al. reported a fetus with agnathia, holoprosencephaly and situs inversus. İn our case, the full term infant had facial anomalies only[9].

Most severe HPE cases and cyclopia can be diagnosed by US during early antenatal care[10]. Early prenatal diagnosis of this malformation along with genetic counseling usually leads to the termination of pregnancy. The medical laws in our country permit the termination of pregnancy if major congenital abnormalities are detected and the parents give consent. However, in this case, the patient was not registered for antenatal care in our hospital until labor pain began. Hence neither US, nor fetal MRI nor cytogenetic analysis was performed earlier, and diagnosis could not be made until labor had begun. Furthermore, the mom suffered the great psychological pain of delivering a living baby with serious deformities, who died soon after birth.

Alobar HPE with cyclopia is a rare, lethal congenital anomaly. The prognosis of HPE varies depending on the type of disease and is related to the presence of extracerebral abnormalities. The diagnosis and awareness of the spectrum of sonographic findings of alobar HPE, cyclopia and other abnormalities by early US are important for early termination of pregnancy to minimize the physiological and psychological impact of such anomalies on the mother and family.

Conflictof interests:

None declared.

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