



True Cyclopia and Proboscis: an Interesting Prenatal Condition with Normal Nose

Prenatal Olarak Tanısı Konulmuş, Normal Burunu Olan Gerçek Siklopi ve Probosis Olgusu

True Cyclopia and Proboscis

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Özet

Siklopi iki gözün orta hatta tek bir orbita içinde birleştiği yüzün anatomik bir deformitesidir. Çok nadir olarak görülen bu durum ölü doğumlar dâhil yaklaşık 100.000 doğumda bir görülmektedir. Bildiğimiz kadarıyla literatürde gerçek siklopi, probosis, holoprozensefali ile birlikte normal burun görünümü olan bir rapor bulunmamaktadır. Biz bu olgu sunumumuzda 2. trimesterde, tek göz küreli, probosis olan ve normal burun görünümlü bir olguyu paylaşacağız.

Anahtar Kelimeler

Gerçek Siklopi; Proposis; Holoprozensefali; Prenatal Tanı

Abstract

Cyclopia is a deformation of the facial skeleton with one eye orbit formed in the place where both eyes should be present. Its rate is almost 1 in 100,000 births including stillbirths are identified as cyclopean. To our knowledge a cyclopean which is including true cyclopia, proboscis, HPE, but normal nose has been never reported. In this current study we describe a second trimester fetus with, single bony orbit, proboscis; but normal nose.

Keywords

True Cyclopia; Proboscis; Holoprosencephalia; Prenatal Diagnosis

DOI: 10.4328/JCAM.1588

Received: 21.01.2013 Accepted: 15.02.2013 Printed: 01.07.2016

J Clin Anal Med 2016;7(4): 542-4

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Introduction

Cyclopia is an extreme and interesting fetal malformation which includes a single palpebral fissure as well as proboscis associated with severe brain malformations. Its rate of occurrence is almost 1 in 100,000 births, including stillbirths identified as cyclopean [1].

The ethmoid complex, rooted in the prechordal mesoderm, plays an important role in the development of the midline and symmetry of the fetal face. Flaws in the development of the ethmoid complex lead to severe malformations of the whole, middle or upper parts of the face. In situations without ethmoid process a structure, called the proboscis, develops above the eyes. This structure is a hollow tube made of cartilage coated with respiratory epithelium. Histologic findings show the proboscis is a similar structure to the nose, which includes respiratory epithelium. This indicates that it results from a problem during development of the frontal region of the nose.

In English-language literature 257 cyclopic cases have been reported until now and 81 of these cases had chromosomal abnormalities. The combination of true cyclopia, proboscis and holoprosencephalia (HPE) is an extremely rare condition [1]. All cases of this combination were reported with the absence of a nose. To our knowledge this case is the first which includes true cyclopia, proboscis, and HPE, but with a normal nose.

Additionally when we look at the literature; most of the images and reports relate to stillbirth or postpartum fetuses' which had cyclopia and proboscis. In this report we present a prenatal diagnosis of true cyclopia, proboscis, but normal nose in an 18-week fetus by 2-D, 3-D and 4-D imaging.

Case Report

A 33-year-old, gravida 3, para 2 woman attended for 2nd trimester fetal "anatomic survey" sonographic scanning at 18+4/7 weeks of gestation. Her obstetrical history revealed spontaneous birth of a male fetus of 3120 g and a Cesarean section female fetus of 4200 g at term gestation, without any anomalies. The parents were not consanguineous and they had no dysmorphic features or congenital anomalies in their family history. There was no history of using an intrauterine device for contraception or drug abuse during early pregnancy and serological screening for TORCH was negative.

On detailed ultrasound examination an abnormal image of the facial structures and fetal brain was seen. Two-dimensional (2-D) ultrasound findings were; an alobar holoprocencephaly with facial anomalies including hypotelorism, fusion of the orbits, single eye and proboscis (Video 1, Figure1). On axial cranial ultrasound examination; thalamic fusion, single cerebrum and mono-ventricle were seen (Figure 2). In 3-D real time imaging and on the coronal profile view of the fetal face a single bony orbit and the proboscis above it was seen as a snout like protrusion from the face (Video 2, Figure 3). There was no abnormality on sonographic examination of other parts of the fetus. Amniocentesis was performed and normal fetal karyotype (46,XX) was diagnosed. After receiving counseling the parents decided to terminate the pregnancy on the basis of the ultrasound abnormalities, but they refused an autopsy. Postmortem physical examination confirmed 280 gr female fetus with cyclopia and proboscis as well as normal nose. Other parts of the body had no additional anomalies.



Figure 1. The 2-D ultrasound image of axial plan of cranium that revealed proboscis and a single bony orbit

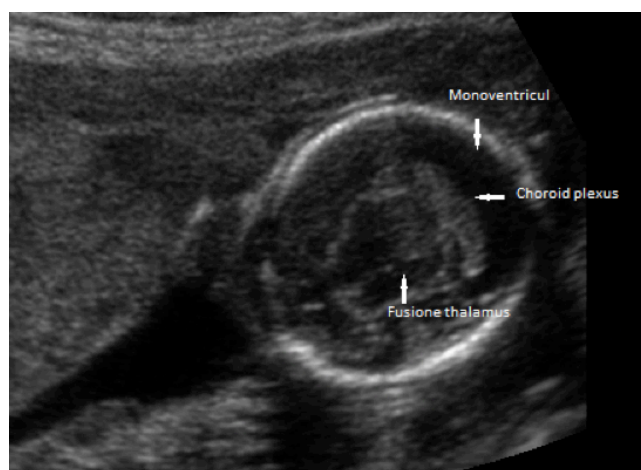


Figure 2. Axial plan of cranium; thalamic fusion and mono-ventricle

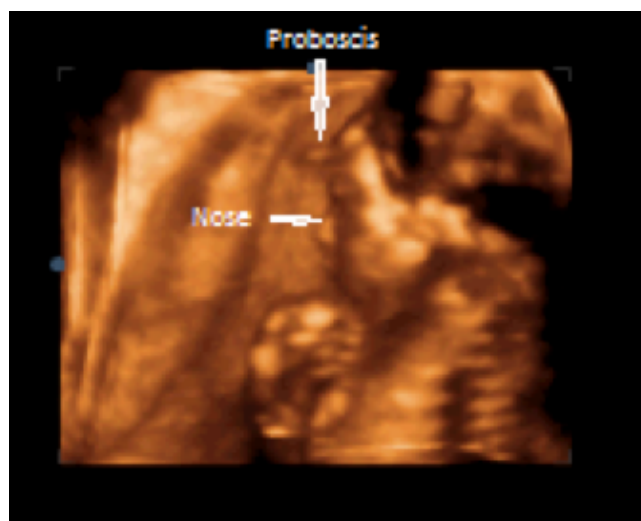


Figure 3. The 3-D ultrasound image that revealed proboscis and a normal nose

Discussion

True cyclopia is a rare anomaly in which the organogenetic development of two separate eyes is suppressed and is associated with severe brain malformations. In cases of true cyclopia with normal karyotype and nose, a demonstration of HPE using 2-D, 3-D and real time imaging has never been reported to our knowledge. Our case report offers these insights. It is characterized by a serious median faciocerebral development deformity

and craniofacial abnormalities, such as mild microcephaly with a single central incisor [2].

The absence of the facial midline bones above the maxilla and the presence of a proboscis as a nose-like structure above the cyclopic eye both mean that there is a developmental defect in the fronto-nasal facial process of this fetus. In this case we have an important question; it may be possible, but we don't know, that cyclopia could deform the nasal structure in the later period of pregnancy [4].

The orbital region is grossly deformed, resulting in the formation of a central cavity 'pseudo orbit', with absence of the nasal cavity and presence of a rudimentary proboscis above the pseudo-orbit. If two globes are found with a different degree of fusion in the pseudo-orbit, the condition is called synophthalmos. A much rarer anomaly is true cyclopia, wherein only one eye is present. Underlying brain malformation is usually alobar holoprosencephaly with microcephaly. It cannot be treated or cured. The brain is so radically reduced and simplified that there is no way to "repair" it and it is incompatible with life. The etiology is not well known. Suggested risk factors include chromosomal abnormalities, potentially teratogenic environmental factors during organogenesis, infections during pregnancy (TORCH), or drugs taken during pregnancy (alcohol, aspirin, lithium, anticonvulsants, hormones, retinoic acid, anticancer agents and fertility drugs) have all been regarded as the basis of this anomaly [1,2,3]. In this case there were no potential risk factors or genetic disorder.

If the facial defects are not easily detected when the fetal position is suboptimal during the 2-D sonographic imaging, 3-D or 4-D scan may play an important role in confirming such defects. Additionally 3-D and 4-D sonography of the facial features can provide more convincing evidence for prenatal diagnosis and counseling. Consequently; true cyclopia is an extremely rare fetal malformation that can be accompanied and confused by proboscis and normal nose.

Competing interests

The authors declare that they have no competing interests.

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How to cite this article:

Uysal F, Uysal A, Güngör AÇ, Coşar E. True Cyclopia and Proboscis: an Interesting Prenatal Condition with Normal Nose. *J Clin Anal Med* 2016;7(4): 542-4.