

Fasial Anomali ve Siklopinin Eşlik Ettiği Holoprozensefali: Olgu Sunumu
Holoprosencephaly Associated with Facial Anomaly and Cyclopia: A Case Report
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Özet

Holoprozensefali tam olarak bölünmeyen serebral hemisferler sonucunda anormal ya da eksik beyin yapılarının görüldüğü bir anomalidir. Doğumlarda prevalansı 1/16.000 olarak bildirilmiştir. Çoğu fetusta siklopi, hipotelorizm, probosus varlığı, nasal kemik yokluğu, yarı damak ve yarı dudak gibi fasial anomaliler de mevcuttur. En ciddi formu alobar holoprozensefalidir. Özellikle Trizomi 13 en sık olmak üzere yaklaşık yarısında kromozomal anomali mevcuttur. Prognoz oldukça kötü olup ya intrauterin ya da doğumdan hemen sonra fetal ölüm görülür. Biz bu olgu sunumunda terminasyonu kabul etmeyen 37. gebelik haftasındaki fasiyal anomali ve siklopiyanın eşlik ettiği holoprozensefali vakasını sunmayı amaçladık.

Anahtar Kelimeler: Holoprosensefali, alobar, anomali, siklopi

Abstract

Holoprosencephaly is an anomaly in which abnormal or missing brain structures are seen as a result of cerebral hemispheres that are not fully divided. The prevalence at birth was reported as 1/16,000. Most fetuses also have facial anomalies such as cyclopia, hypotelorism, proboscis, absence of nasal bone, cleft palate, and cleft lip. The most serious form is alobar holoprosencephaly. Chromosomal anomalies are present in about half of the cases, especially Trisomy 13. Prognosis is poor and fetal death occurs either intrauterine or immediately after birth. In this case report, we aimed to present a case of holoprosencephaly with facial anomaly and cyclopia at 37th gestational week which does not accept termination.

Key Words: Holoprosencephaly, alobar, abnormality, cyclopia.

Introduction

Holoprosencephaly is an anomaly in which abnormal or missing brain structures are seen and caused by cerebral hemispheres that are not fully divisible. The prevalence was reported as 1/16000 (1,2). Facial anomalies such as cyclopia, hypotelorism, proboscis, absence of nasal bone, cleft lip and cleft lip are present in most fetuses⁽²⁾. Holoprosencephaly occurs during fetal development due to lack of prosencephalon division at 4 and 8 gestational weeks. There are four subtype antagonisms according to the degree of forebrain development; alobar, semilobar, lobar and middle interhemispheric combination variants (3). The most serious form is alobar holoprosencephaly. Chromosomal anomalies are present in about half of the cases, especially Trisomy 13. Prognosis is poor and fetal death occurs either intrauterine or shortly after birth. In this case report, we aimed to present a case of holoprosencephaly accompanied by facial anomaly and cyclopian at the 37th gestational week which does not accept termination.

Case Report

A 22-year-old gravida 1, according to the latest menstrual period, a 37 gestational week pregnant woman applied to our

hospital due to premature ruptures of membranes. The vaginal examination of the patient revealed that 2 cm dilatation and 70% effacement in the cervix and also active amniotic fluid appearance. One alive fetus was seen in the breech presentation, where there was a heart beating with alobar holoprosencephaly anomaly in the ultrasonic examination. With the current findings, the patient was admitted to the obstetrics service. There was no virtue according to resume or family history in the patient's anamnesis. During pregnancy, she was informed that she regularly had her follow-ups, but always applied to different centers. It was learned that the baby was found to be anomalous at the 4th month of pregnancy and the termination was suggested, but the patient did not accept the termination. Cesarean scheduled for pregnancy with active delivery because of prolonged labor and breech presentation. 1950 grams weight of a newborn girl was born with caesarean section as 1st minute APGAR score: 2 and 5th minute score APGAR: 0. Newborn fetal cardiopulmonary resuscitation (CPR) was performed on account of a heart rate under 60 beat/minute. The resuscitation was not received any answer for about 20 minutes and the baby was admitted to the exitus. Physical examination of the postmortem newborn revealed severe facial anomaly (cyclopia,

rudimentary nasal mucosa and incomplete structure). The baby had no nasal or oral orifices and there was a cyclopia (Figure-1). In the first planet, it was considered Trizomi 13. But postnatal pathological examination was not possible because the family did not accept the autopsy. The mother was discharged on the 2nd day of hospitalization without any postoperative complications. The present case report has been prepared with the Helsinki Declaration principles in mind and with the permission of the family upon publication of the case.

Figure 1. Holoprosencephaly associated with facial anomaly and cyclopia



Discussion:

Holoprosencephaly is a rare congenital anomaly and the incidence of birth is one in 16,000 live births (1).

Holoprosencephaly occurs during fetal development due to lack of prosencephalon division at 4 and 8 gestational weeks (2). There are four sub-types of holoprosencephaly according to the degree of development of forebrain and the most severe form is alobar (1,4). There is no interhemispheric fissure formed because of uncomplete association of structures belonging to Proencephalus, only one ventricle is existed. The development rate of the brain determines the shape of the face of the fetus.

Holoprosencephaly can occur with environmental or genetic causes, and holoprosencephaly is most commonly associated with trisomy 13 (2,4,5). The prognosis of Holoprosencephaly is quite poor, and the survey differs in general from the subtypes, to the degree of holoprosencephaly and to the percentage of dysmorphic condition in the face. In the literature only 50% of infants with alobar holoprosencephaly have lived up to the 4th month of the first year of life and 20% of them have been followed up for 12 months (3-5). Early diagnosis of holoprosencephaly and termination of pregnancy should be the first step to be taken because of a fatal outcome for fetus and protection the mother from complications related pregnancy and labor. In our case, Holoprosencephaly was diagnosed at the 16th gestastional weeks.

Although termination was recommended as a treatment option to the family but termination was not accepted by family. The fetus was delivered with a cesarean section due to breech presentation and died after birth when the heart rate fell below 60 beats/minute at 5 minutes and did not respond to resuscitation for 20 minutes.

In conclusion, holoprosencephaly is an anomaly rarely seen and fatal to the fetus in generally. Early determination of this anomaly and the need to take early steps to terminate the fetus may lead to avoid risks of pregnancy and labor. Also it may be acceptable for the family to terminate the pregnancy that alobar holoprosencephalic baby will not survive.

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