Case Report

Alobar Holoprosencephaly

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SUMMARY
ALOBAR HOLOPROSENCEPHALY
Background: Alobar holoprosencephaly is characterised by median malformations of the face and brain due to incomplete cleavage and morphogenesis of the forebrain. The alobar holoprosencephaly occurs in about 0.6 to 1.9 of 10.000 deliveries. Median facial abnormalities consist of orbital hypotelorism, cyclopia in combination with a flat nose or proboscis and oral deformities can be diagnosed with ultrasonography at the early antenatal period.
Observation: In this case report, we introduced a fetus with an alobar holoprosencephaly diagnosed at 24 weeks of gestation according to BPD and FL measurements. In obstetric ultrasonography, spinal cord, vertebras and spinal canal were normal in structure, but interhemispheric fissure, falx cerebri and corpus callosum were not visualised. There was single-chamber ventricle in semihiron shape that continued with a dorsal cyst filling the calvarium. Decision for pregnancy termination was made after the consultation with the pediatricians. In neonatal examination, there was no gross deformity of the trunk and extremities. As a facial deformity, cyclopia and 3x2 cm sized proboscis attacked above the insignificant nasal structure was detected and only one eye was present at the middle of the face.
Key Words: Alobar holoprosencephaly, Prenatal diagnosis

ÖZET
ALOBAR HOLOPROSENSEFALİ
Amaç: Alobar holoprosensefali ön beyinin inkomplet bölünme ve morfogenezinden kaynaklanan yüzün orta hat malformasyonları ile karakterize bir anomalidir. 10 bin doğumda 0.6-1.9 oranında görülmektedir. Alobar holoprosensefalide görülen orta hat yüz anomalileri olan orbital hipotelorizm, silik burun veya probosis ile birlikte siklopia ve oral deformiteler gebeliğin erken dönemlerinde ultrasonografik olarak tespit edilebilmektedir.
Olgu: Bu vaka takdiminde BPD ve FL ölçümlerine göre 24. gebelik haftasında bir alobar holoprosensefali olgusu sunuldu. Obstetrik ultrasonografik incelemede, spinal kanal, vertebralar ve kord normal olarak izlenirken, kranyumda interhemisferik fissür, falks serebri ve korpus kallosum izlenmedi. 3. ventrikül saptanmayıp monoventrikül hali mevcuttu. Yarımay şeklindeki holoventrikül büyük bir dorsal kist ile devam etmekte ve kalvaryumu doldurmakta idi. İstenen pediatri konsültasyonu sonucu indüksiyon uygulanarak gebelik sonlandırıldı. Yapılan neonatal incelemede gövde ve ekstremitelerde belirgin bir deformite tespit edilmedi. Frontal bölgede 3x2 cm'lik yumuşak doku tespit edildi. Yarımay yapılar silik olup, yüzde orta hatta tek göz mevcuttu.
Anahtar Kelimeler: Alobar holoprosensefali, Prenatal tanı

The incidence of alobar holoprosencephaly is 0.6-1.9/10.000 in deliveries. Alobar holoprosencephaly is composed of anomalies that result from interruption of the development of anterior brain at the early period of embryonic life. It is characterised by unvisualized intracranial midline structures with only one central ventricle wide in size (2,3). Besides that, there is cyclopia and hypotelorism (1). The prognosis of these fetuses are poor and they die in intrauterin life or immeadiately after delivery (1,4). In this case report, we introduced a fetus with an alobar holoprosencephaly diagnosed at antenatal period and terminated by the willing of family.

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CASE

The pregnant woman was aged 28 years old with gravity 2 and parity 1. Her husbands age was 30 years old. There was no family history of significant medical problem in both parents. In mother's obstetric history, there was a caesarean section 3 years ago because of cephalo pelvic disproportion, delivered 3000 g healthy infant without

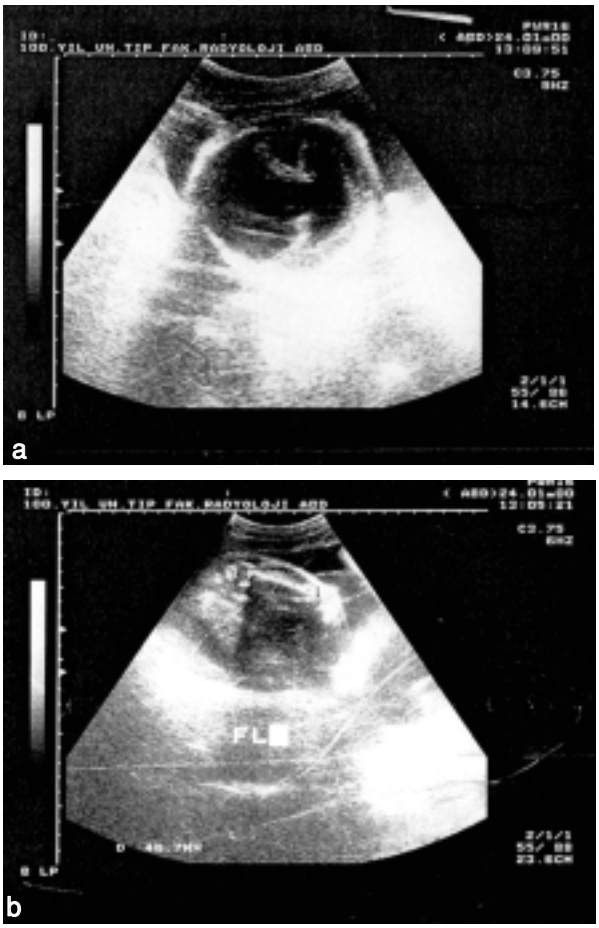

Fig. 1A- 1B. A 24 weeks old pregnancy according to BPD and FL. Inter-hemispheric fissure, falx cerebri, corpus callosum and third ventricle are not visible and there is only one ventricle, semilunar shaped holoventricle, continuing with a dorsal cyst that fills the calvarium.

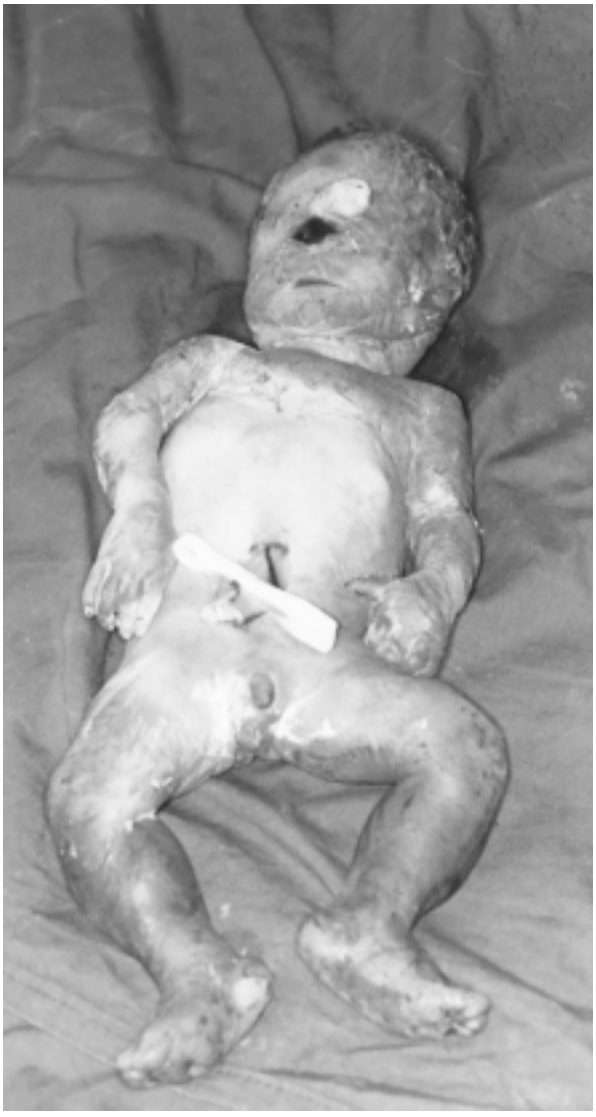

Fig. 2. There is no gross deformity in the trunk and extremities

any detected anomaly. In this pregnancy, she recieved no antenatal care until 28 weeks 2 days of gestational age according to her last normal menstrual period. Her first visit was to our out patient department and as a component of routine examination, on obstetric ultrasonography according to BPD and FL 24 weeks old pregnancy was detected (fig. 1A- 1B). Spinal cord, vertebras and spinal canal were normal but inter-hemispheric fissure, falx cerebri, corpus callosum and third ventricle could not be visualised and there was only one ventricle, semilunar shaped holoventricle, continued with a dorsal cyst that filled the calvarium (fig. 1A). These findings are relevant to alobar holoprosencephaly. After consultating with the pediatricians, decision for termination of the pregnancy was made. Family was informed and they agreed with the decision. By labor induction, a male fetus with 3 apgar score at 1st minute weighing 1400 gr was delivered and in his neonatal examination, there was no gross deformity in the trunk and extremities (fig. 2). At the frontal region of the head, 3x2 cm sized, pedinculated soft tissue was detected. Nasal structures were insignificant and only one eye was present at the middle of the face (fig. 3). Fetus died 5 minutes after the delivery.

DISCUSSION

Craniospinal anomalies are the most commonly seen congenital anomalies and can be diagnosed easily by ultrasonographic imaging. Ultrasonographic visualisation of fetus during pregnancy can detect large group of pathologies at the early period of embriyonal life. The perfect time for that is at the 16th to 18th weeks of pregnancy. The major

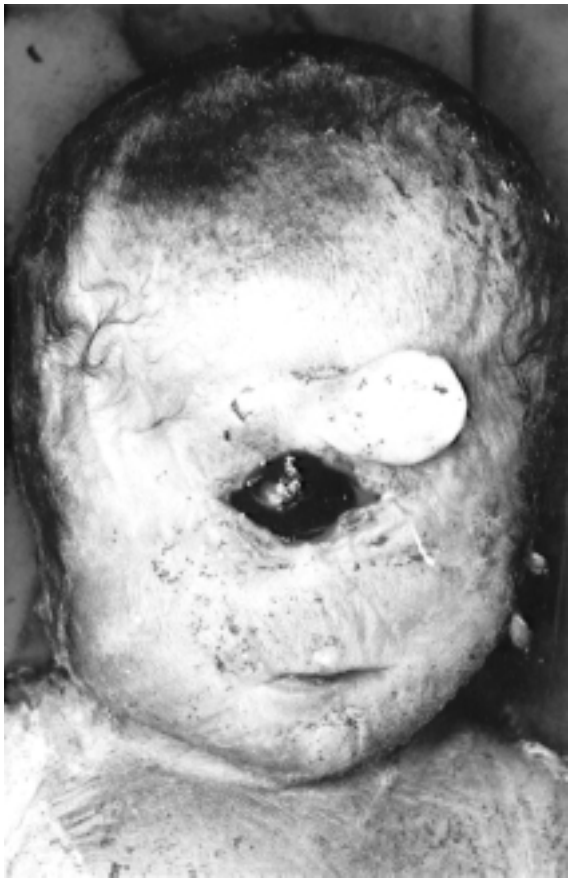

Fig. 3. 3x2 cm sized, pedinculated soft tissue with insignificant nasal structures and only one eye is present in the middle of the face.

congenital anomalies can be easily diagnosed by performing ultrasonography at that time interval. More detailed ultrasonographic imaging is necessary for lethal diagnosis. In this way, pregnancy can be terminated at an early period of gestational age in cases of lethal malformations.

Holoprosencephaly can be divided into labor, semi-lobar and alobar subgroups according to cleavage defect of prosencephalon during developmental period. Among them, the most easily diagnosed anomaly is the alobar holoprosencephaly which is characterised by unvisualised midline intracranial structures, fusion of thalamus and presence of only one ventricle wide in size (2,3). Brain is small in size. Instead of ventricular system, only lateral and third ventricles are present and there is no connection between them, the monoventricle system in relation with dorsal sac is present. Thalamus and corpus striatum are in connection. Corpus callosum, fornices, falx cerebri, optic tractus and olfactory prominence are not present midbrain, brain stem and cerebellum are normal in structures. There is cyclopia and hypotelorism (1,6). In semilobar holoprosencephaly, falx cerebri and temporal lobe are developed partially in posterior part but there is thalamic and ventricular fusion in anterior part. In lobar holoprosencephaly, only lateral ventricules are present and continue with the other anterior portion (2,3).

Facial anomalies are present together with holoprosencephaly and its progression is severe. The severe forms of facial dimorphism are generally seen in alobar holoprosencephaly. Cyclopia and ethmocephaly are always present with alobar holoprosencephaly (1,7). In our case, there was a 3x4 cm sized pedinculated soft tissue located at the frontal region with insignificant nasal structures. And one eye was present in the middle of the face.

Fetal karyotyping must be regarded in the presence of all CNS anomalies. The specific craniofacial findings for trisomy 13 are cleft palate and lip, hypotelorism, depressed rudimentary nose and cyclopia. Karyotyping is absolutely indicated in alobar holoprosencephaly cases because high incidence of chromosome anomalies have been detected. Trisomy 13 is frequently seen but other anomalies can also be detected (1,7). Unfortunately we could not perform karyotyping because of technical difficulties.

In intrauterine life, all the variations of holoprosencephaly can be diagnosed by ultrasonographic imaging. Detection of severe forms are more common but also lobar holoprosencephaly can be detected (4,5,8). Obstetric approach for holoprosencephaly cases is dependent upon the gestational age and severity of anomaly at the time of diagnosis (9). If diagnosis of holoprosencephaly can be made before 24th weeks of gestation, parents can prefer termination of pregnancy. Development of macrocephaly because of ventricular dilatation can prevent vaginal delivery, so that in hopeless patients, cephalosynthesis can be performed to avoid cesarean section. Recently some cases are presented from our country remarking bad fetal prognosis and need to terminate pregnancy (10,11).

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