CASE REPORT

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Fetal Anomaly Which is Exencephaly and Accompanying Omphalocele at First Trimester

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ABSTRACT Exencephaly is a rare anomaly in which the brain tissue goes out of cranium from skull. It can be easily diagnosed with ultrasonography in early period. Omphalocele is the defect of the abdominal wall covered with by membrane on abdominal midline. Omphalocele sac is included often the liver with small intestines; rarely spleen, stomach, bladder, uterus and ovaries. Other system anomalies can be accompany to the omphalocele at 60%. A 34-years-old female, exencephaly and accompanying omphalocele was diagnosed with by ultrasonography at the 13th week of pregnancy. Ultrasonographic findings and prognosis of the patient were reported to the family. The family decided to termination of the pregnancy. It is possible to early diagnose congenital cranial anomalies such as exencephaly by ultrasonography. In this case, we present to discuss a patient who had an omphaloce with central nervous system anomaly at the 13th week of pregnancy and terminated pregnancy for this reason.

Keywords: Abnormalities; congenital abnormalities; hernia, umbilical; neural tube defects; pregnancy reduction, multifetal

Exencephaly is a rare anomaly in which the brain tissue goes out of cranium from skull. It can be easily diagnosed with ultrasonography in early period.[1,2] The ossification of the cranial bones starts from the 10 th week of gestation.[3] 9-week 3-days acrania was reported in the literature.[4] Anencephaly; characterized by the absence of cerebral hemispheres in intrauterine life, exencephaly; characterized by large and disorganized brain tissue located outside the cranium, acalvaria; characterized by the absence of bone structures in calvarium, these central nervous system anomalies are the congenital anomalies that should be suppose in differential diagnosis.[5] Omphalocele is the defect of the abdominal wall covered with by membrane on abdominal midline. Omphalocele sac is included often the liver with small intestines; rarely spleen, stomach, bladder, uterus and ovaries.[6,7] Omphalocele can be seen one in 2500-5000 gestations and the rate of comorbidity of other system anomalies in this pathological condition has been determined as 60%.[8] Trisomies 18 fetuses with severe central nervous system anomalies accompanied by the omphalocel were described in literature.[9]

In this case, we present to discuss a patient who had an omphaloce with central nervous system anomaly at the 13th week of pregnancy and terminated pregnancy for this reason.

CASE REPORT

A 34-year-old female patient (G9 P3 A5 Y3) was referred to our clinic for the purpose of detailed ultrasonographic examination because of the head bones of fetuse not observed in an performed ultrasonographic examination of 1st trimester screening of the 13th week of pregnancy at the external center. A single live fetus compatible with 13-weeks 5-day was observed during ultrasonographic examination. Some pathological features were observed during ultrasonographic examination such as; supra orbital calvarial bones of fetuse were not observed and the morphology of brain tissue was preserved (exencephalic), and the internal organs of the abdomen are hernia from the anterior wall of the abdomen (omphalocele). Exencephaly accompanied by omphalocele was diagnosed by ultrasonography. There was no characteristic in the history and family of the case.

Chorionic villus sampling (CVS) was suggested to the family to exclude a possible genetic abnormality. But the family did not allow the CVS. Ultrasonographic findings and prognosis were explaned to the family. The family decided to terminate the pregnancy when the termination option was offered. Therapeutic curettage was applied to patients with misoprostol.

Cranial bone were not observed although brain tissue is normal and also omphalocele was seen in abortion material (Figure 1). Placenta and supplements were sent to pathology. The case was discharged with healing.

DISCUSSION

Acrania is a rare developmental anomaly, one type of neural tube defects (NTD), is characterised by the partial or complete absence of cranial bones despite the development near complete of brain tissue.[10] The exencephaly is a congenital anomaly characterized by disorganized brain tissue in a large amount extending from the abnormal skull.[11] This brain tissue is covered with epithelium and can undergo rapid necrosis by the amniotic fluid and as a result of can become small and degenerate to the anencephaly.[12] The exencephaly is a rare congenital anomaly (spectrum of acrania-exencephaly-anencephaly) that occurs in the spectrum of the acrania and anencephaly.[4,11]

Many factors play a role in the etiology of neural tube defects. Genetic transition was detected in studies when NTD seen with a syndrome. There is usually no family history in the acrania-exencephaly, so chromosomal analysis is not recommended since most of the cases karyotype are normal. The risk of recurrence of NTD increase 10-20 times who have previously in NTD birth stories. In researchs, NTD is frequently seen who feed devoid of zinc and folic acid.[11,13,14]

Previously, the cases of anencephaly diagnosed in the 2nd trimester is diagnosed in the early pregnancy period due to the 1st trimester screening test being between 11th and 14th weeks.[15] The ossification of the cranial bones starts from the 10th week of gestation and this ossification is visible in the ultrasound from the 11th week.[3] 9-week 3-days acrania was reported in the literature. But most of the cases are reported at the earliest in 10-14. gestation weeks.[4] Ultrasonographic findings of first trimester exencephaly; decreased size of the cranial region compared to the chest, irregularity of cranial surface, and amniotic fluid echogenicity.[16] Calvarial bones were not observed in the first trimester ultrasonography of our case but omphalocele and normal brain tissue were observed.

Omphalocel can be seen one in 2500-5000 gestations, is defined an extra embryonic hernia as a result of the termination of ventral medial migration of dermatomyomas.[8] Trisomies, renal, neurological and other gastrointestinal abnormalities can be seen with omphalocele.[17] There were central nervous system anomalies and accompanied exencephalia in our case. The coexistence of the two may be part of some specific syndromes.

Some anomalies can be accompained to the acrania-exencephaly-anencephaly sac as neural tube defects, omphalocel, liver and heart anomalies, cleft palate-lip, microphthalmia etc.[18,19] In this case, we present exencephaly with accompanied omphalocele. It might be a genetic transition, but the family did not approve genetic research.

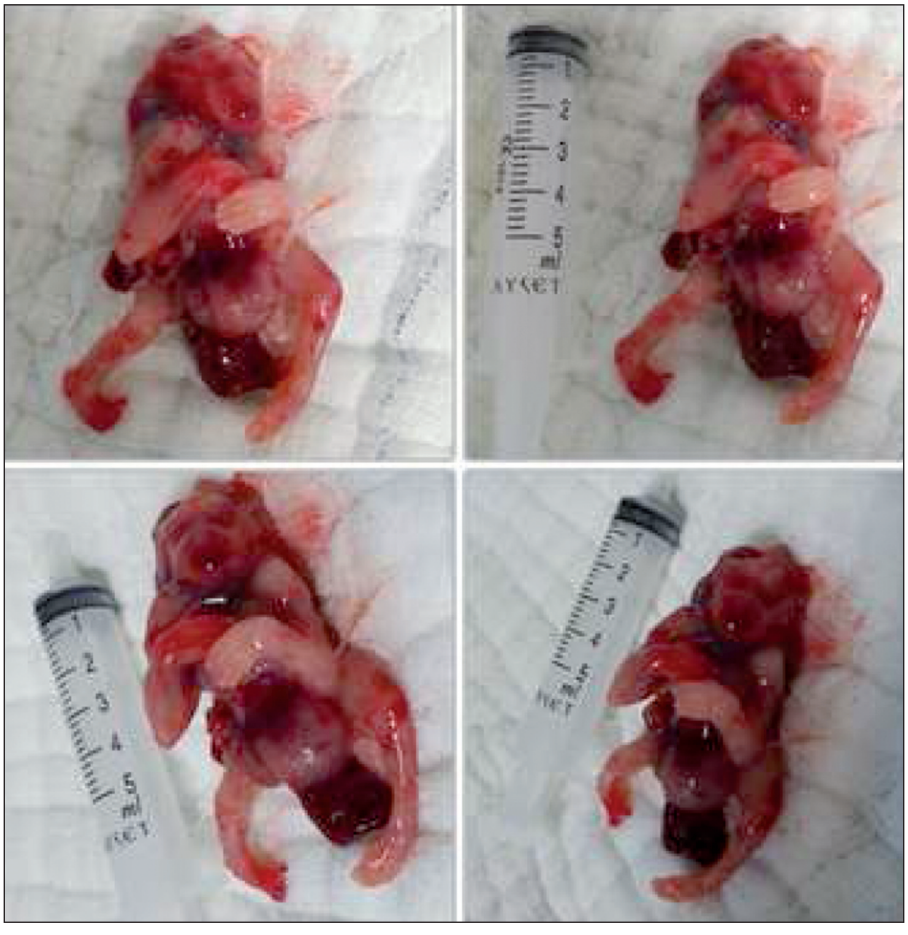

FIGURE 1: Cranial bones were not observed although brain tissue is normal and also omphalocele was seen in abortion material on figure.

It is possible to early diagnose of congenital cranial anomalies by ultrasonography such as exencephalia. It should not be forgotten that it can be together with other congenital anomalies. Exencephaly and also acrania-anencephaly in this spectrum have lethal reasons and also additional anomalies can be accompanying such as omphalocele as in our case. In this reason, may be careful ultrasonographic examination prenatally diagnosed and it will enable families to have early termination rights.

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Conflict of Interest

No conflicts of interest between the authors and / or family members of the scientific and medical committee members or members of the potential conflicts of interest, counseling, expertise, working conditions, share holding and similar situations in any firm.

Authorship Contributions

Idea/Concept: *Erhan Hüseyin Cömert, Hidayet Şal, Süleyman Güven, Emine Seda Güvendağ Güven;* ***Design:*** *Erhan Hüseyin Cömert, Hidayet Şal;* ***Control/Supervision:*** *Süleyman Güven, Emine Seda Güvendağ Güven;* ***Data Collection and/or Processing:*** *Erhan Hüseyin Cömert, Hidayet Şal, Süleyman Güven, Emine Seda Güvendağ Güven;* ***Analysis and/or Interpretation:*** *Erhan Hüseyin Cömert, Hidayet Şal, Süleyman Güven, Emine Seda Güvendağ Güven;* ***Literature Review:*** *Erhan Hüseyin Cömert, Hidayet Şal;* ***Writing the Article:*** *Erhan Hüseyin Cömert, Hidayet Şal;* ***Critical Review:*** *Süleyman Güven, Emine Seda Güvendağ Güven;* ***References and Fundings:*** *Süleyman Güven, Emine Seda Güvendağ Güven;* ***Materials:*** *Erhan Hüseyin Cömert, Hidayet Şal, Süleyman Güven, Emine Seda Güvendağ Güven.*

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