ACRANIA IN TERM FETUSA

Termde fetusta akrani

İLKER KAHRAMANOĞLU, MERVE BAKTIROĞLU, FATMA FERDA VERİT

Süleymaniye Kadın Hastalıkları Ve Doğum,kadın Hastalıkları Ve Doğum,istanbul, Türkiye
Suleymaniye Birth And Women Health Education And Research Hospital, Department Of Obstetrics And Gynecology,istanbul,turkey

ÖZET

Fetal akrani, kraniyal çatı kemiklerinin kısmi veya tam yokluğu ile karakterize, nadir görülen,yaşam şansı çok düşük olan konjenital bir patolojidir. Takipsiz gebelerde ve tanısı konup terminasyon istemeyen hastalarda maternal invazif girişimin ve aileye psikolojik zararın arttığı, maliyetin yükseldiği göz önünde tutulmalıdır.
2. trimesterde fetal akrani tanısı konulup terminasyon önerilen, aile olarak terminasyonu kabul etmeyen, miadında normal doğum yapan gebe sunulmuştur.
Anahtar Kelimeler: Akrani, nöral tüp defekti, term fetus

ABSTRACT

Acrania is a rare fetal malformation, characterized by abnormal development of the calvarium with a large mass of disorganized brain tissue. Early diagnosis of acrania is important for determining the necessity to terminate the pregnancy and also to decrease the negative psychological and financial effects on patients and doctors.

In this report, we present a case of term acrania, diagnosed in second trimester, refused termination because of religious beliefs.

Key words: Acrania, neural tube defect, term fetus

INTRODUCTİON

Neural tube defects cover a broad spectrum of morphologic anomalies of the central nervous system(CNS) .They represent the most common CNS malformations, by having an incidence of just 1–2 per 1000 live births.(1) The most severe of these defects is the sequence of acrania which is an uncommon congenital anomaly characterized with partial or complete absence of the flat skull bones covering the brain and the cerebral hemispheres, while present, are disorganized.(2) Prenatal diagnosis of cases is important in order to terminate the pregnancy as early as possible in order to avoid negative psychological and financial effects on patients. With the improvement of the new techniques, careful inspection of fetal structure using ultrasound during NT measurement at 10–13 weeks of gestation provides an encouraging advantage for early diagnosis of fetal acrania (3, 4). In this report, we present a case of term acrania diagnosed in second trimester, refused termination because of religious beliefs.

CASE REPORT

A 16-year-old, primiparous woman at 38 weeks, menstrual age, was admitted to our clinic because of frequent contractions. Her medical history did not raise any concerns. Family history was negative for congenital and chromosomal anomalies. Acrania was detected in the second trimester (21 weeks, menstrual age); pregnancy termination was offered but patient was refused because of religious beliefs and continued to have regular visits to different hospitals.

In our physical examination, 6 cm cervical dilataion and %80 effacement was detected. In ultrasound examination, fetus compatible with 29 weeks' gestation, an absence of crania vault, smaller brain tissue, vertebral defect extend to sacrum was revealed. Amniotic fluid index was measured as 400 mm, means polyhydroamnios. An informed consent form was obtained from the patient.

After an hour, patient had normal delivery. The baby's weight was 1460 gr, Apgar scores were 0 and 1 at 1 and 5 min, respectively. She was apneic and hypotonic, and was placed on mechanical ventilation. After 60 minutes, the baby died. Macroscopic evaluation was revealed that cranial bones were absent, brain tissue was atrophic and was only covered by a thin membrane, defect in vertebral column extends to sacrum was seen(figure 1, 2)

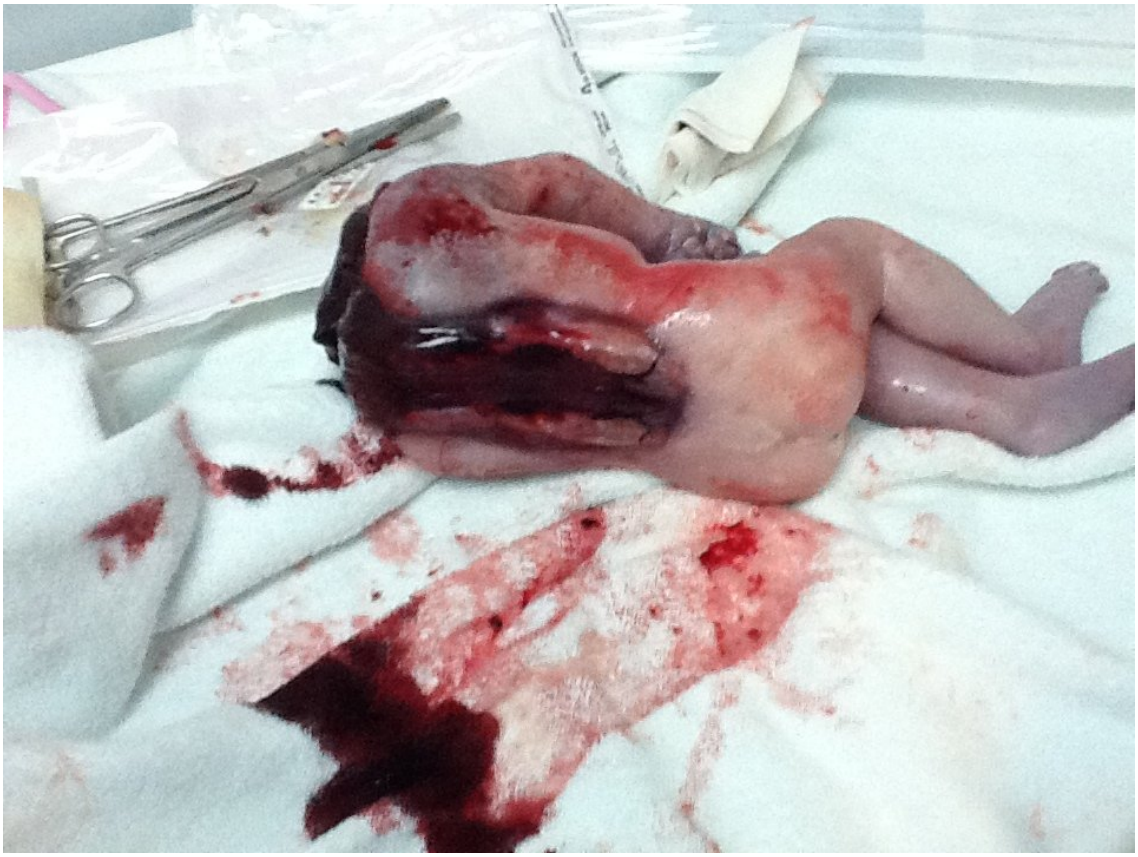

Figure 1 Photograph of the whole body taken after delivery. Posterior view of fetus showed a gross spinal defect.

Figure 2. There is obvious absence of normal calvaria.

The mother was psyhologically affected and after discharged, she was refferred to a psychiatric clinic.

DİSCUSSİON

Fetal acrania is a rare congenital anomaly resulting from failure of the mesenchyma to migrate
under the ectoderm overlying the brain tissue to form the bone tissue over the cerebral hemispheres.
This abnormality occurs at the beginning of the 4th week of embryonic development, when the anterior neuropore closes. The brain tissue is covered only by a thin membrane and is therefore exposed to the amniotic fluid. The cerebral hemispheres, although present are anatomically disorganized(2, 3). The fetal cranium is not fully calcified before 10-11 weeks; therefore, a first trimester diagnosis must be made with caution(5). Although fetuses with acrania are reported to have more echogenic amniotic fluids which would be a sign that can lead to early diagnosis during the first trimester (10), acrania can be diagnosed with transabdominal and transvaginal US during the end of the 1st trimester or beginning of the 2nd trimester.

In sonographic examination related diseases include anencephaly, large cephalocele, osteogenesis imperfecta, and hypophosphatasia. These should be kept in mind in order to make differential diagnosis(6). Under ultrasonography, both of the exposed hemispheres of the brain are only covered by a thin membrane called

the ‘Mickey-Mouse’ sign. Most cases of acrania eventually progress to anencephaly and the features frequently seen in cases with bulging eyes called the ‘frog-eye’ sign, which is easily diagnosed during the second trimester(2).

As it is a lethal anomaly, early ultrasound diagnosis enables patients to create a timely termination of the pregnancy(3). Then, it would be possible to reduce the financial disadvantages and worse psychologic effect on both the clinicians and the parents. After a certain diagnosis of fetal acrania is made, patients should be informed properly that it’s inviable condition for prevention of term pregnancy(7). Folate or vitamin B9 supplementation is also recommended to reduce the risk of neural tube defects and other congenital abnormalities like cardiovascular diseases, cleft lip and palate, urogenital abnormalities and limb reductions in her future pregnancies (8). Furthermore,fetuses with an neural tube defects and other congenital abnormalities have a significantly increased risk for low birth weight(9) and in order to prevent life threatening neonatal morbidity such as asphyxia**,** sepsis, hypoglycemia etc in these situations , we have to prevent neural tube defects with necessary folate or vitamin B9 supplements.

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