"Acrania": Two Case Reports With Prenatal Ultrasound Results

"Akrani": Prenatal Ultrasonografi Bulguları ile İki Olgu Sunumu

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Abstract
Acrania is a rare congenital anomaly and characterized by partial or complete absence of the calvarium with abnormal brain tissue development. The pathogenesis of acrania is unknown and differential diagnosis should be searched to rule out other similer conditions to anencephaly and acalvaria. Diagnosis of cranial bone defects can be established by ultrasonography in the first trimester of pregnancy. We report two cases, diagnosed prenatally by ultrasonography, one with isolated acrania and the other one associated with meningocele.

Key Words: **Acrania; Prenatal diagnosis; Ultrasonography, Prenatal.**

Özet
Akrani kraniyal kemiklerin bir kısmının ya da tamamının yokluğu ve eşlik eden anormal beyin dokusu gelişimi ile karakterli nadir bir konjenital anomalidir. Akraninin patogenezi bilinmemektedir. Kraniyal kemik defektlerinin tanısı gebeliğin ilk trimesterinde ultrasonografi ile yapılabilmektedir. Biz bu yazıda prenatal dönemde tanı konulan izole akrani ve akraniye eşlik eden meningosel olgularının ultrasonografi bulgularını sunduk.

Anahtar Kelimeler: **Akrani; Prenatal Ultrasonografi; Prenatal tanı.**

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Introduction

Fetal acrania is an uncommon congenital anomaly characterized with partial or complete absence of the calvarium (1). Prenatal diagnosis and close monitoring of cases which have a minimal chance of survival in the postnatal period, bears importance (2). Currently, cranial bone defects can be determined in the first trimester by transabdominal and transvaginal ultrasonography (US) (3).

In the present report, we reported US results of isolated acrania cases which were diagnosed on 15th and 16th weeks by transabdominal US, and the associated meningocele finding.

Case Report

Both cases had healthy familial histories including no consanguineous marriage and teratogenic drug usage, or any known systemic disease.

Case 1. Transabdominal US examination performed on the 28-year-old pregnant woman who was estimated to be 15 weeks pregnant according to her last date of menstruation, revealed a single and alive fetus. Examination of the cranium exhibited a proportionately small head compared to the body; cerebral structure was present, however, bony structure of the cranium displayed no echogenicity (Figure 1a, b). No accompanying malformation was observed in the systemic screen.

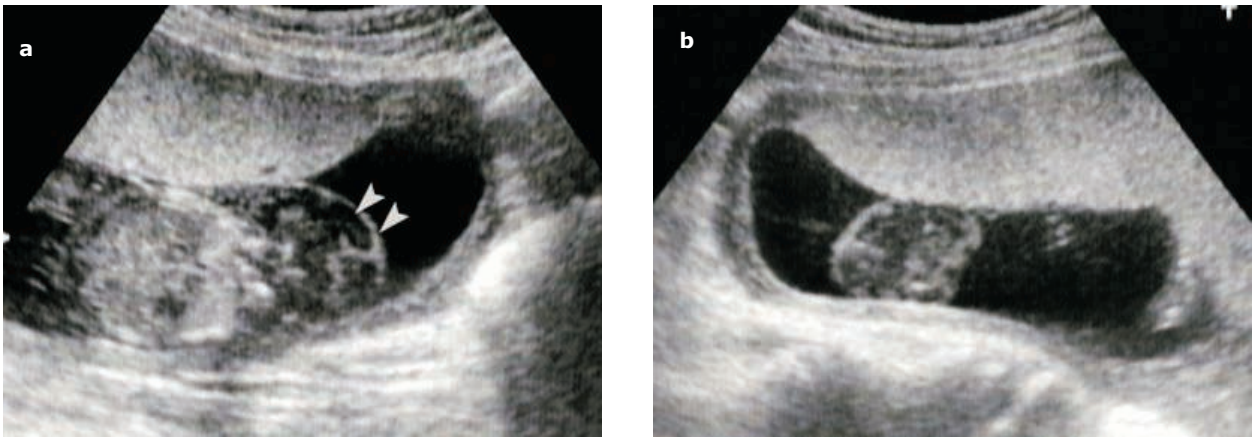

Figure 1. While brain parenchyma is observed in the longitudinal (a) and axial (b) abdominal USs, there is no calvarial bone structure (arrow heads).

Case 2. Transabdominal US examination performed on the 22-year-old pregnant woman who was estimated to be 16 weeks pregnant according to her last date of menstruation, revealed similar results, and moreover, exhibited a cystic lesion of 15x11mm size showing regular contour pattern and anechoic feature in the cervicothoracic region suggestive of meningocele.

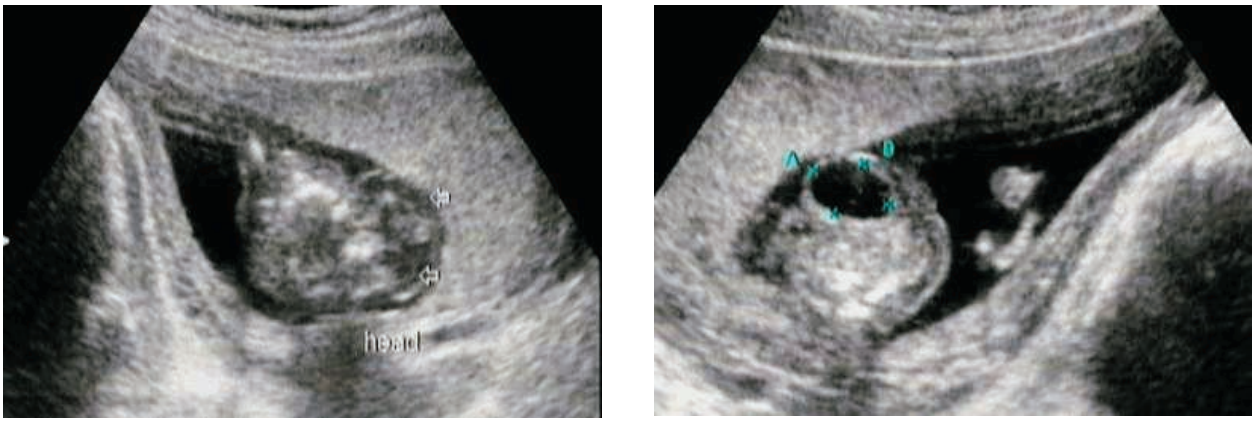

Figure 2. Transabdominal US examination of the 16-weeks pregnant patient, displaying a smaller head disproportionate to the body, alongside absence of echogenicity of the cranial bones.

Resim 3. İkinci olguda servikotorasik bölgede, anekoik, düzgün konturlu, kistik lezyon izlenmekte.

Medical abortion was performed on the cases and the eventual examination revealed cerebral hemispheres that were enclosed in a thin membrane, no development of cranial bones, and a meningocele in the cervicothoracic region accompanying acrania in the 2nd case.

Discussion

Acrania is an uncommon developmental anomaly which is characterized with complete or partial absence of cranium despite almost normal development of brain tissue (1). The most important theory aiming to explain the mechanism of fetal acrania development, suggests failure of mesenchymal migration during the 4th development week of embryonic life as the underlying cause (4).

Fetal acrania cases may be associated with central nervous system defects such as neural tube defect, cervicothoracic spina bifida (5), omphalocele, liver and heart abnormalities, foot deformities and anomalies such as microphtalmia (4), and more uncommonly amniotic band syndrome (6). In the present report, our 2nd case had a meningocele localized in the cervicothoracic region, accompanying acrania.

Because cranium ossification is seen by the 13th week, US diagnosis is recommended to be established after 11-12 weeks (7). In the present report, diagnoses are achieved by the 15th and 16th weeks. US diagnosis is reached by determining a head disproportionately smaller than the body, and absence of calvarial bones despite presence of cortical brain tissue and facial bones (3,8). Both our cases had the described findings.

Differential diagnosis should include anencephaly, exencephaly, and acalvaria (9). Anencephaly is a developmental anomaly in which major portion of the brain is not developed and defects in cranium are observed. While exencephaly is characterized with acrania and the resultant protrusion of disorganized brain tissue, acalvaria is described by absence of calvarial bones and cerebral hemispheres despite normal development of condocranium (1) .

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.

Because acrania is a fatal pathology, medical abortion should be performed immediately upon diagnosis (3). Early diagnosis and ending of the pregnancy at an early period bears importance because it reduces interventions that could be used during the last trimester, presents a lower cost, and decreases the amount of psychologic effects that can be encountered both in the physician and the family (11).

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