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CASE REPORT

Ultrasonographic prenatal diagnosis of isolated acephaly

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ABSTRACT
First trimester obstetric ultrasonography of a 32-year-old female patient revealed a 13-week-old (according to the length of the femur and abdominal circumference) intrauterine live pregnancy with the absence of the fetal head. Medical abortus was performed with the diagnosis of acephaly and final diagnosis was confirmed by pathological and radiological examinations. We present and discuss the possible etiopathologic mechanisms of an acephaly case, which is described as acardia-acephaly complex in the literature, and usually appears in cases of twin reversed arterial perfusion sequence, but has not been previously reported as an isolated finding.

Key words: • *acephaly* • *prenatal diagnosis* • *ultrasonography* • *amniotic band syndrome*

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Central nervous system (CNS) malformations constitute a broad range of congenital anomalies (1–4). Almost all of them result in severe and irreversible neurological defects, and many cannot support life. Ultrasonography (US) is an indispensable imaging method for diagnosing and managing these malformations intrauterinely. Among CNS malformations, neural tube defects are the most common, in which very rare and severe anomalies like acephaly can also be seen (1). In this case report, we present an isolated acephaly diagnosed during a first trimester US examination, discuss the possible etiopathologic mechanisms, and review the related literature.

Case report

A 32-year-old female patient was referred to the obstetrics and gynecology clinic of our hospital for routine pregnancy follow-up. Her history was unremarkable, including the lack of teratogen exposure or infections. Pedigree analysis indicated that the mother and father were first-degree step-cousins. Their first pregnancy produced a daughter that died at 11 months because of a posterior auricular tumor of unknown pathology. Their second and third pregnancies ended with spontaneous abortus in the 10th gestational week, and 2 subsequent pregnancies produced 2 healthy children (a 10-year-old girl and an 8-year-old boy).

During the current first trimester follow-up, the couple was referred to the radiology department for obstetric US. The obstetric US revealed a 13-week-old in utero live fetus, according to fetal abdominal circumference (66 mm) and femur length (11 mm). Fetal cardiac activity was positive (180/min) and fetal somatic movements existed; however, the fetal cranium and its supplements were not present (Fig. 1). The small amount of amniotic fluid present was striking. In the light of these findings the case was diagnosed as acephaly. Medical abortus was performed 2 days after the US diagnosis. No maternal complications appeared following the procedure. Assessment of the abortus material revealed an ex-fetus body and bilateral extremities with male genitalia, and as revealed by US, the fetal neck and head were not observed (Fig. 2). To evaluate possible bone defects, post-mortem direct X-ray images were taken. Although all bony structures were normal and symmetrically present, the 3rd cervical vertebra and higher cervical or cranial bone structures were absent (Fig. 3). Autopsy examination showed properly placed and anatomically normal thoracic and abdominal internal organs, without any anomalies. The fetal heart and urinary system were particularly normal. Pathological sections were taken from the defective cervical region and microscopic evaluations revealed no evidence of bone or neural tissue that might be related to the fetal cranium. Considering all the findings, the diagnosis of acephaly was con-

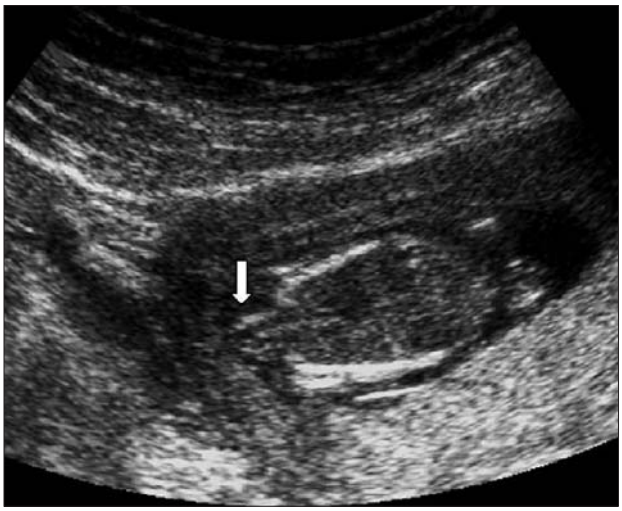

Figure 1. Fetus is seen in the coronal plane on transverse transabdominal US image. The lack of the fetal head beyond the 3rd cervical vertebra *(arrow)* is striking.

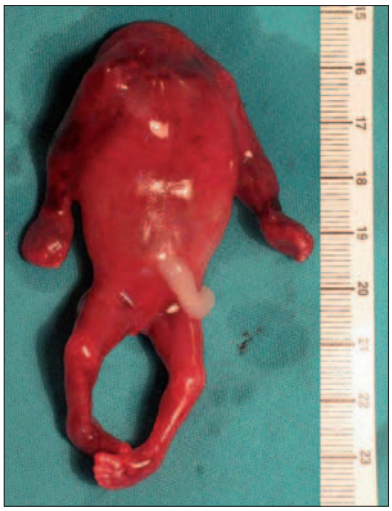

Figure 2. Front macroscopic view of the ex-fetus. Fetal head cannot be seen.

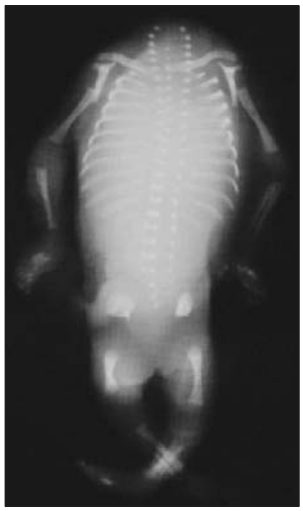

Figure 3. Post-mortem anteroposterior direct X-ray. Fetal cranial bone structures are not visible.

firmed. Blood samples of the parents were examined at the medical genetics department and no chromosomal anomalies were detected.

Discussion

Acephaly is the complete lack of a head and is one of the severe fetal anomalies that do not support life. In a large study by Pinar et al. (1) 4,122 case autopsies of CNS anomalies were reviewed for a period of over 37 years and only 4 acephaly cases were reported. Acephaly, which is an extremely rare anomaly, is believed to mostly accompany complicated monochorionic-monoamniotic twin pregnancies, when twin reversed arterial perfusion (TRAP) sequence occurs with the presence of acardia (5–7). In this condition, usually one of the twins is normal and, with the help of in utero interventions, can survive (8, 9). On the other hand, the other sibling presents with acephaly and acardia, as well as various pulmonary and genitourinary anomalies. Environmental, rather than genetic, factors are implicated in the etiopathogenesis as only one sibling is affected (7). Our case differs from others as it was a singleton pregnancy, and, to the best of our knowledge, it is the first isolated acephaly case detected. In addition, the lack of any chromosomal disorder in the parents and no additional fetal anomalies in the post-mortem examinations led us to consider that environmental factors, rather than genetic problems, were the cause.

Amniotic band syndrome (ABS), which can be the cause of many defects of the fetal head, trunk, and extremities due to mechanical pressure, is the first etiologic factor to be considered in the formation of acephaly (10–19). The prevalence of ABS among live births is in the range of 1/1,200–1/15,000 (20). As different defects occur and 3 different germ layer-derived tissues suffer in various manners, ABS is thought to arise due to mechanical factors, such as premature amniotic rupture, rather than genetic factors (21). ABS, which usually presents sporadically, rarely results from teratogen agent exposure or hereditary connective tissue diseases (Ehlers-Danlos type IV syndrome, osteogenesis imperfecta, etc.) (22). ABS can often result in mild to moderate anomalies and amputations of the trunk and extremities, or craniofacial defects like cleft palate, which can be functionally corrected by orthopedic or reconstructive surgery. Rarer severe anomalies, such as acrania, acalvaria, exencephaly, anencephaly, and, as in the presented case, acephaly, may arise, all of which do not support life. As ABS may affect only one part of the body, there are situations in which more than one amniotic band may cause multiple organ deformities. In the presented case, poor obstetric history of the mother and low levels of amniotic fluid support the diagnosis of ABS in the formation of acephaly.

US is more helpful in assessing fetal anomalies caused by ABS than ABS itself. The literature indicates that it is not always possible to show the amniotic bands in ABS cases (15, 23, 24). US examinations to scan fetal anomalies are often carried out in the second trimester, between the 18th and 22nd gestational week (25). In order to detect severe anomalies earlier and to terminate the pregnancy when necessary, a first trimester US examination that is performed between the 11th and 14th weeks for double test becomes more important in fetal anomaly scanning.

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