An Unusual First-Trimester Ultrasound Presentation of the Acrania-Anencephaly Sequence

The "Turkish Turban" Sign

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A previously unrecognized first-trimester presentation of the acrania-anencephaly sequence is described. Ultrasound features included a constriction ring around the external base of the developing skull and an enlarged globular head, resembling a Turkish turban, with large cystic spaces replacing the brain. This constellation of findings was noted in 3 first-trimester fetuses. In 2 of them, it was possible to identify the amniotic membrane attached to the constriction ring. One case presented with anencephaly and fetal demise at 16 weeks. The other 2 women terminated the pregnancies and aborted anencephalic fetuses. This subtype of the acrania-anencephaly sequence could represent an earlier segmental rupture of the amnion, which subsequently entraps the developing fetal skull.

Key Words—acrania; acrania-anencephaly sequence; amniotic band syndrome; first-trimester ultrasound; open neural tube defect; prenatal diagnosis

The acrania-anencephaly sequence, also known as the exencephaly-anencephaly sequence,[1] is a lethal neural tube defect (NTD) characterized by a sequence of events triggered by nondevelopment of the mesenchymal coverage of the brain (future calvarian bones). The unprotected brain tissue subsequently undergoes progressive destruction and degeneration after mechanical and chemical trauma, ultimately resulting in absent cerebral hemispheres.[1] Although the condition of anencephaly has been recognized for centuries,[2] fetal acrania was only recognized as a clinical entity in 1982.[3] This was as a result of incorporating ultrasound (US) into antenatal care, thus making it possible to identify absent calvarian bones in association with a substantial amount of preserved brain tissue in early pregnancy.[3] Subsequent prenatal US studies have demonstrated that fetal acrania/exencephaly is the predecessor of anencephaly, as this condition typically leads to the classic presentation of anencephaly in the second and third trimesters of pregnancy or at the time of delivery.[4–6]

The acrania-anencephaly sequence is now a well-recognized condition that can be diagnosed with confidence by using US during early pregnancy.[7] The main features include absent calvarian bones in association with differing amounts of disorganized brain tissue.[7] An associated but indirect sign of acrania-anencephaly is the presence of echogenic amniotic fluid,[8] resulting from bleeding and disaggregation of dysplastic brain tissue.[5,6] Recent efforts to classify the different types of acrania-anencephaly sequence when presenting in the first trimester

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Abbreviations

NTD, neural tube defect; US, ultrasound

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have been reported.[9] These include the exposed cerebral hemispheres taking on a bilobular shape (the "Mickey Mouse" sign),[9,10] as well as cystic, foreshortened, elongated, and overriding types of acrania.[9] The aim of this report is to describe another subtype that we have termed the "Turkish turban" type. We also provide US evidence that this presentation is probably secondary to early-stage amniotic band syndrome, which suggests that this specific type of NTD is the result of a disruptive insult.

Methods and Results

All women participating in this study attended our centers voluntarily and consented to the US examination, including the use of transvaginal and 3-dimensional US technology. The protocol for the first-trimester US examination was approved by the local Institutional Review Boards. Due to the deidentified retrospective nature of the study, permission from the Ethics Committee was not deemed necessary.

The main features of the first-trimester US in 3 cases of acrania-anencephaly sequence are described. Two patients were referred after abnormal US findings were noted by their attending obstetricians. In the other, the abnormal findings were detected during the routine 11- to 13-week scan. All 3 cases shared similar characteristics in terms of the aspect of the fetal head. Figures 1–3 show the main 2- and 3-dimensional US findings in these cases. In every case, the head was molded by a constriction ring formed around the external base of the developing skull, without involving any of the face or the rest of the fetal body. The head was globularly enlarged and covered by a membrane with no evidence of calvarian bones. The brain was replaced by several large cystic spaces, while some remnants of brain tissue were identified over the base of the skull. In 2 of the 3 cases, a clear connection between the constriction ring and the amniotic membrane was shown (Figures 1 and 2). This suggested that the amnion was wrapped around the base of the skull, thus producing the constriction ring. No additional anomalies were detected, and all fetuses had a normal nuchal translucency thickness. One of the fetuses had bradycardia of 130 beats per minute at the time of the evaluation, well below the fifth percentile for gestational age according to the nomogram of Hyett et al.[11] Another had reversed flow velocity waveforms in the ductus venosus during atrial contraction. A follow-up scan was only available in 1 of the cases and confirmed a rupturing of the cranial sac, leading to early second-trimester anencephaly. This woman arrived at the emergency department with vaginal bleeding, with US showing intrauterine demise. The pregnancies were terminated in the 2 remaining cases, which led to the abortion of anencephalic fetuses after complete disruption of the membrane covering the head.

Figure 1. Case 1. **A–C**, Transabdominal (**A**) and transvaginal (**B** and **C**) 2-dimensional US views at 13 weeks show an abnormal bulging at the level of the fetal head. No calvarian bones are identified, and the brain is replaced by several cystic spaces. Note the amniotic membrane attached to the abnormal head (large arrow) seen in **C**. **D**, Surface-rendered 3-dimensional US view of the fetal head (**D**). Note the Turkish turban appearance of the fetal head. Small arrows in **C** and **D** denote the constriction ring around the external base of the developing skull.

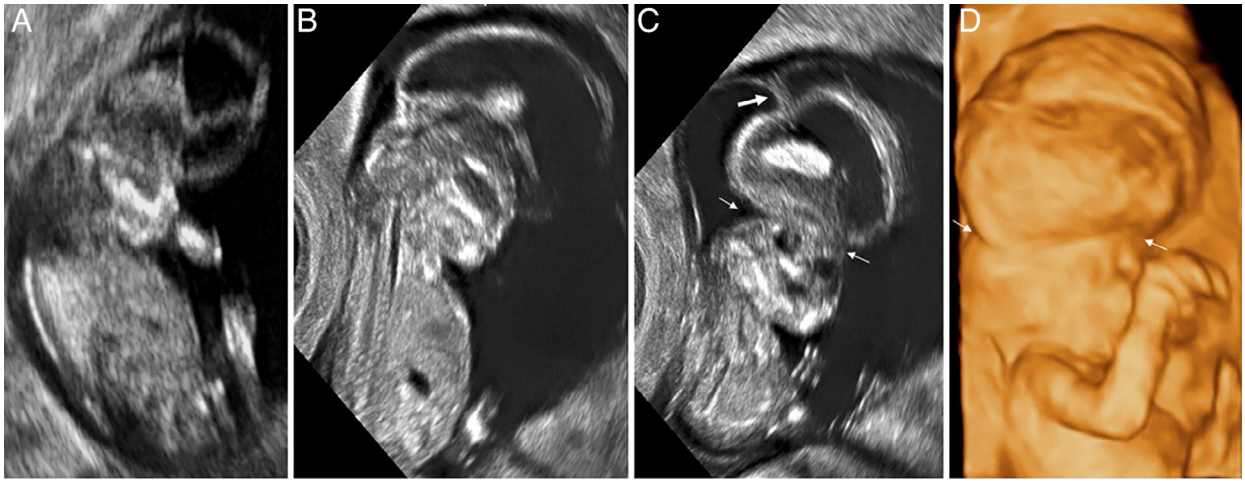


Figure 2. Case 2. **A** and **B**, Two-dimensional (**A**) and 3-dimensional (**B**) US views of the fetal head at 13 weeks. Note the constriction ring around the external base of the developing skull, the enlarged globular head, and cystic spaces replacing the brain. The amniotic membrane attached to the fetal head is denoted by arrows.

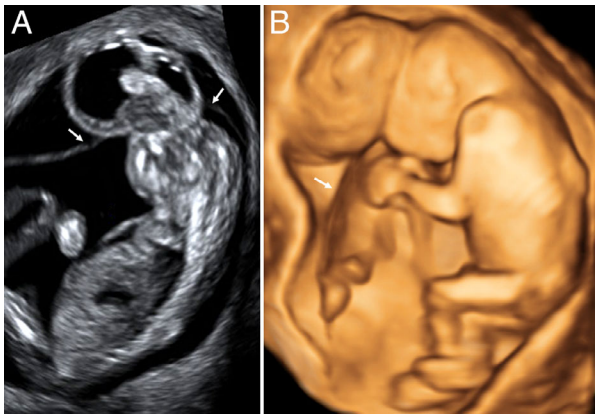
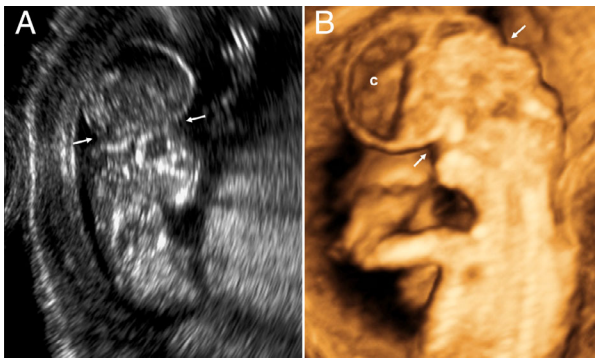


Figure 3. Case 3. **A** and **B**, Two-dimensional (**A**) and 3-dimensional (**B**) US views of the fetal head at 12 weeks. Note the constriction ring around the external base of the developing skull (arrows), the enlarged globular head, and the cystic spaces (c) replacing the brain.



Discussion

The etiology of anencephaly is multifactorial.[12–14] The main etiopathogenic mechanisms include open NTDs as a result of the anterior neuropore failing to close around the fourth week of development, as well as disruptive conditions such as amniotic band syndrome.[12–14] Strictly speaking, the latter condition is not a primary open NTD. However, it may lead to similar morphologic appearances at the time of the delivery. The cases of acrania-anencephaly sequence that we report seemed to be the result of amniotic bands that were specifically attached to the external base of the developing skull.

A number of different types of the acrania-anencephaly sequence have been previously described in the literature.[7,9,10] However, none of these descriptions matched the US and clinical characteristics observed in our cases. Indeed, our 3 cases presented a constriction ring around the external base of the developing fetal skull, with a detailed assessment of this area revealing that the amniotic membrane was wrapped around the head in at least 2 of them. In the other remaining case, a focused examination to rule out amniotic bands was not conducted, as the operator was unaware of this presentation and, in retrospect, it was not specifically searched for. We speculate that during the progressive growth of the fetal head, the constriction ring compresses the base of the skull, leading to impaired circulation to the vulnerable developing brain as well as abnormal circulation of the cerebrospinal fluid. This ultimately leads to an enlarged globular head due to accumulation of cerebrospinal fluid within the head, absent calvarian bones, and cystic degeneration of the brain.

In conclusion, we have described a previously unreported presentation of the acrania-anencephaly sequence in the first trimester that is probably the result of amniotic bands forming around the external base of the developing skull. Sonologists should be aware of this presentation, which is often due to a disruptive process. As such, this condition is probably not dependent on the folate pathway. This is an important finding, highlighting the fact that every effort should be made to establish the etiology of acrania. This is particularly relevant, as the risk of recurrence in these cases must be considered much less than acrania resulting from a genetic mutation of the folate pathway. Accordingly, these patients can be reassured of a much lower risk of recurrence than the 5% recurrence risk associated with primary open NTDs.[12–14]

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