

Case report

Fetal Acrania: A Case Report

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SUMMARY

Introduction. Acrania is a very rare lethal congenital malformation characterized by an absence of the cranial vault with developed cerebral hemispheres. It is sometimes confused with anencephaly in which both the forebrain and neurocranium are not developed. Although these two conditions principally differ in their morphology and pathogenesis, acrania may be a precursor of the development of anencephaly through the process known as Acrania-exencephaly-anencephaly sequence.

Case report. A 27-year-old woman was diagnosed to have a viable fetus with an absent skull vault and uncovered brain directly exposed to amniotic cavity. A diagnosis of acrania was made. A medical abortion was performed in the 14th week of pregnancy. A gross examination of the formalin-fixed fetus revealed a complete absence of the calva and brain tissue. The cranial structures stop abruptly above the orbits and the fetus showed a triangular face with bulging eyes. At the top of the head, a huge defect with a residual thin covering membrane was visible. According to the gross morphology and in correlation with previous ultrasound findings, the pathologist established the diagnosis of secondary anencephaly.

Conclusion. This paper suggests that many of the reported anencephaly cases that are diagnosed during the second or third trimester of pregnancy might represent the end of a spectrum that initially appears as isolated acrania. This may be the reason for much higher global incidence of anencephaly compared to acrania.

Keywords: acrania, exencephaly, anencephaly, acalvaria

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INTRODUCTION

Fetal acrania is a very rare congenital anomaly, in which there is a partial (meroacrania) or complete (holoacrania) absence of the flat skull bones with developed, albeit abnormal brain hemispheres identified in direct contact with amniotic fluid (1 - 4). It is sometimes confused with another severe malformation of the neural tube – anencephaly, in which both the forebrain and neurocranium are not developed (5 - 6). Although these two entities principally differ in their morphology and pathogenesis, acrania may be a precursor of the development of anencephaly through the process known as acrania-exencephaly-anencephaly sequence (AEAS). The AEAS refers to the progression from an absent cranial vault (acrania) showing relatively normal-appearing exposed brain (exencephaly) detected in the first trimester to no recognisable brain parenchymal tissue (secondary anencephaly) found in the later stages of pregnancy (3, 4, 7 - 10). In the present paper, a case of fetal acrania is described with demonstration of prenatal sonographic and postmortem pathological findings.

CASE REPORT

A 27-year-old pregnant woman, gravida 2, para 1, was diagnosed to have a single fetus with a severe somatic anomaly. Her medical history revealed she had given birth to a child with tetralogy of Fallot two years before. No genetic disorder in her family had been documented. An ultrasound scan performed in the 13th week of her second pregnancy showed a living fetus with a completely absent skull vault. The brain was developed and directly exposed to amniotic cavity (Figure 1 and 2). The facial components of the head were identified. A diagnosis of acrania was stated. No other apparent somatic aberrations were detected. Amniotic fluid volume was adequate (normohydramnios). As the fetal anomaly was considered incompatible with life, the mother decided to terminate pregnancy. The abortion was induced in the 14th week of gestation per vias naturales and resulted in a macerated dead fetus (crown-rump length 60 mm, weight 22 g.) of indeterminate gender. After the baby was born, the placenta was delivered (weight 60 g.). Both were

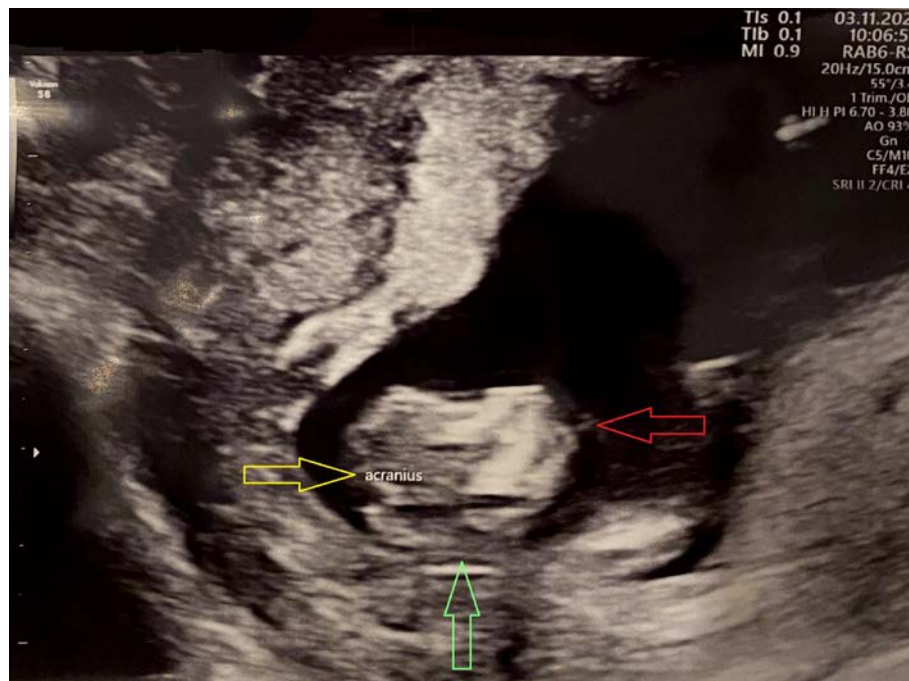


Figure 1. Grey scale ultrasound image showing the absence of the skull vault with unprotected brain, suggestive of acrania with exencephaly (yellow arrow – brain, red arrow – face, green arrow – occiput)

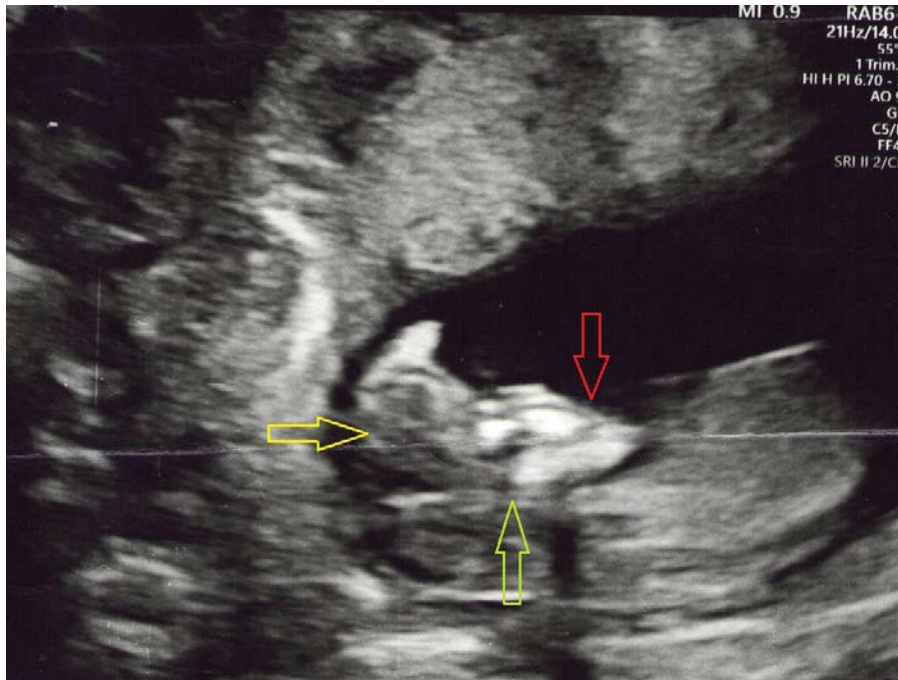


Figure 2. Another grey scale ultrasound scan showing lack of the calvarium with brain matter herniating out (yellow arrow – brain, red arrow – face, green arrow – occiput)



Figure 3. The appearance of the triangular face with bulging eyes. The neurocranium is absent (after fixation in formalin)



Figure 4. A detail on the huge pathologic opening at the top of the head (after fixation in formalin)



Figure 5. Sagittal section revealed neither the calva nor the brain (after fixation in formalin)

fixed in formalin and sent for histological investigation. The hospitalisation period was uneventful, and the patient was discharged in a good condition.

A gross examination of the formalin-fixed fetus revealed a complete absence of the calva and brain. The cranial structures stop abruptly above the orbits and the fetus showed a triangular face with bulging eyes („frog-like“ appearance) (Figure 3). At the top of the head, a huge defect with a residual thin covering membrane was visible (Figure 4). After a sagittal incision, neither the calvarial bones nor the brain tissue was detected (Figure 5). The spine was macroscopically without any defect and the underlying skin was intact. Major visceral organs within the thoracic and abdominal cavity were situated in their anatomical positions. Overall microscopic assessment of taken organs was partly limited due to tissue autolysis, but no convincing histopathological changes seemed to be present. According to the gross morphology and in the correlation with previous ultrasound findings, the pathologist established the diagnosis of secondary anencephaly. No information was known regarding a genetic testing of parents.

DISCUSSION

With today's ultrasound technology, it is easy to identify almost all cases of acrania at the end of the first trimester of pregnancy (3, 8, 10). Because acrania is a lethal condition, the vast majority of prenatally diagnosed cases end in a legal termination of pregnancy for medical reasons. That is why this entity can be encountered in pathological practice mainly in the form of fetal abortion. The pathogenesis of isolated acrania is proposed to be an abnormal migration of the primordial mesenchymal tissue that normally covers the cerebral hemispheres (3, 4, 10). This leads to non-development of calvarial bones, dura mater, and the scalp, while the brain is covered only by a thin, amniotic membrane (10).

The main conditions that must be considered in prenatal ultrasound differential diagnosis are anencephaly and acalvaria. As mentioned previously, anencephaly is a type of the open neural tube defect that results from failed closure of the cranial end of the neural tube (rostral neuropore) during the third to fourth week of embryogenesis (5, 6). The affected babies lack cerebral hemispheres, meninges, bones, and skin (2, 5, 6). Acalvaria, by definition, is the absence of the flat bones and dura mater

of the cranial vault, while the brain is directly covered by skin (2).

As noted above, acrania could transform into anencephaly through the AEAS. According to this theory, the cerebral tissue that is not protected by meninges, cranium and skin undergoes progressive destruction in utero due to exposure to the harmful effect of acidic amniotic fluid (increased urea concentration in the amniotic fluid) and mechanical irritation (3, 4, 7 - 10). This leads to complete or almost complete disappearance of the brain tissue from the 14th week after conception onward (7). The AEAS theory allows understanding the much more frequent occurrence of anencephaly than acrania (10). However, the precise analyzing the AEAS harbors several pitfalls. First of all, the study of the sonographic appearance of sequelae that occurs during the AEAS would require a follow-up of the pregnant women with fetal acrania. Whereas most of them terminate the pregnancy shortly after the first diagnosis, direct observation of this sequence is no longer possible. Nevertheless, certain sonographic features are considered the warning signs for early stage of the AEAS. They include the „beret“ sign, transformation of the „Mickey mouse“ sign into the „frog-eye“ sign, and a slightly increased echogenicity of amniotic fluid (due to free-floating particles of exfoliated neural tissue) (7, 10). From a prognostic point of view, there is no fundamental difference between acrania and anencephaly because both are uniformly lethal. On the other hand, primary anencephaly, as with neural tube defects in general, shows a stronger genetic predisposition. The estimated recurrence rate is 2 - 5% after one affected child (6) and 13% after two affected children (5). In comparison, there is no clear evidence for a specific genetic contribution or increased risk of recurrence in future pregnancies for isolated acrania (1, 2).

CONCLUSION

This paper demonstrates a unique case of the fetus with isolated acrania established by prenatal ultrasound that showed morphological features of secondary anencephaly at postmortem examination. Many of the documented anencephaly cases that are diagnosed during the second or third trimester of pregnancy might represent the end of a spectrum that initially appears as isolated acrania. This may be the reason for much higher reported incidence of fetal anencephaly compared to acrania.

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Fetalna akranija: prikaz slučaja

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SAŽETAK

Uvod. Akranija je veoma retka letalna kongenitalna malformacija koja se karakteriše odsustvom svoda lobanje sa razvijenim cerebralnim hemisferama. Ponekad se greškom dijagnostifikuje kao anencefalija, kod koje i prednji mozak i neurokranijum nisu razvijeni. Premda se ova dva stanja razlikuju u svojoj morfologiji i patogenezi, akranija može prethoditi razvoju anencefalije kroz proces koji se zove akranija-egzencefalija-anencefalija sled.

Prikaz slučaja. Kod dvadesetsedmogodišnje žene dijagnostikovao je održiv fetus sa odsutnim svodom lobanje i nepokrivenim mozgom koji je bio direktno izložen amnionskoj šupljini. Postavljena je dijagnoza akranije. Medicinski abortus urađen je u 14. nedelji trudnoće. Makroskopski pregled fetusa, fiksiranog u formalinu, otkrio je potpuno odsustvo kalvarije i moždanog tkiva. Kranijalne strukture se završavaju odjednom, iznad orbita. Kod fetusa je utvrđen trouglasti oblik lica sa ispupčenim očima. Na vrhu glave uočen je veliki defekt sa rezidualnom tankom membranom. Na osnovu makroskopskog pregleda i prethodnih ultrazvučnih nalaza, patolog je postavio dijagnozu sekundarne anencefalije.

Zaključak. Ovaj rad ukazuje da mnogi od opisanih slučajeva anencefalije koji su dijagnostikovani tokom drugog i trećeg trimestra trudnoće mogu predstavljati kraj spektra koji se inicijalno pojavljuje kao izolovana akranija. Ovo može biti razlog mnogo veće incidencije anencefalije na globalnom nivou u poređenju sa akranijom.

Ključne reči: akranija, egzencefalija, anencefalija, akalvarija