

Acrania: a case report

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Abstract

Acrania is a rare lethal congenital anomaly characterised by an absence of the calvarium. Ultrasound allows early diagnosis of this anomaly. Although acrania associated with anencephaly is a well recognized entity with an incidence of about 10:10,000 births, isolated acrania is a rare anomaly, and its incidence is unknown. We report a 25 year old female patient referred to our ultrasound department for antenatal scan. The fetus was found to have a completely formed brain, base of the skull and facial structures but lacking a cranium.

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INTRODUCTION

Acrania is a developmental anomaly characterized by partial or complete absence of the cranium with complete but abnormal development of brain tissue. It is the product of an ectodermal and mesodermal aberration and occurs after neural tube closure. Thus, acrania differs from anencephaly, which results from a neural tube defect and is characterized by absence or near absence of the cerebral hemispheres and, consequently, the overlying calvaria. The diagnosis of acrania can be established sonographically even in the first trimester if a large mass of disorganized brain tissue covered only by a thin membrane is detected.

CASE REPORT

A 25 year old female was referred to our ultrasound department. Pedigree analysis revealed a history of first degree consanguinity between the parents. Family history was non-corroborative. There was no history of intake of teratogenic drugs and other relevant past illness. Ultrasonography revealed a single alive fetus of 14 weeks gestational age in cephalic presentation with longitudinal lie. Fetal brain was well formed without cranium (acrania). Brain convolutions, inter hemispheric fissure and sulci were clearly identified. A thin membranous structure was covering the brain. The fetal brain appeared to be floating in the amniotic fluid over the base of skull and was easily compressible on probe pressure due to lack of cranium. Increased amount of amniotic fluid was seen. The fetus showed normal cardiac activity (134 beats/min) with normal rhythm. The movements of the fetal body parts were normal and well coordinated. Facial structures were normal. Both orbits were symmetrical in size and shape. Cervical, thoracic and lumbar spine appeared normal with normal morphology of the spinal canal. No mass was seen attached or arising from the spine. Fetal long bones were normal. Placenta was anterior in position and was of normal thickness. The fetus was therapeutically aborted. We correlated our antenatal sonographic findings with gross pathological features.



Figure 1: Fetal Acrania. Gray scale ultrasound of the fetal brain in coronal plane. Gestational age 14 weeks 1day. No skull is seen around the brain.



Figure 2: Fetal Acrania. Gray scale ultrasound of the fetal brain in axial and coronal plane show absent skull bones.

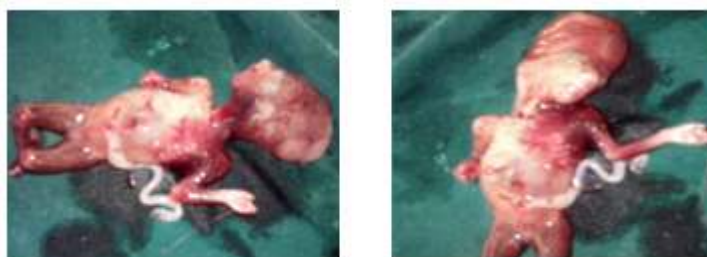


Figure 3: Gross images of the aborted fetus showing absence of skull bones and scalp with exposed brain parenchyma.

DISCUSSION

Fetal acrania (exencephaly) is a congenital abnormality characterized by the complete or partial absence of skull bones surrounding the fetal brain. Lack of mesenchymal migration in the fourth week of embryological age is a proposed mechanism^{1,2}. The fetal cranium is not fully calcified before 10–11 weeks; therefore, a first-trimester diagnosis must be made with caution³. Fetal acrania can be diagnosed from 11 weeks onward. Misdiagnosis may occur if only midsagittal views of the fetus are obtained at 11–14 weeks gestation because the majority of cranial ossification is in the lateral aspects of the frontal bones and lower parietal bones, and no vault ossification is visible in the midline on a perfect midsagittal image. The absence of cranial ossification may not be noted, and the head may appear relatively normal. It is important to look specifically for frontal bone ossification in the axial and coronal planes^{4,6}. Meroacrania refers to absence of the cranium with the exception of the occipital bone. Acrania exposes the brain to the amniotic fluid with risk of friction with uterine wall, placenta and fetal parts, this condition is called exencephaly. In this circumstance, the

unprotected brain tissue undergoes progressive destruction and degeneration due to mechanical and chemical trauma, leading to complete or almost complete disappearance of the brain from 14 weeks onward. The damaged brain tissue can be seen on ultrasound as echogenic particles in the amniotic fluid⁷. 3-D US may contribute to early detection of fetal acrania and provide a novel visual depiction of this defect after reconstruction⁵. The differential diagnosis includes anencephaly and large cephaloceles. In anencephaly, cerebral tissue is completely absent; while in cephaloceles, the cranial vault can always be detected and a part of the brain is intracranial. A distinction should also be made between acrania and conditions characterized by lack of mineralization of the skull bones such as hypophosphatasia and osteogenesis imperfecta Type II. In these skeletal dysplasias, the intracranial anatomy is normal, and the brain is surrounded by a thick layer of tissue representing soft tissues and unossified bone. Bowing, fractures or shortening of long bones are usually present and knowledge of family history aid in differential diagnosis¹.

CONCLUSION

Fetal acrania is a rare and lethal congenital anomaly that warrants the identification of fetal skull and cranium around the brain that should be normally calcified⁸. Antenatal identification allows the clinician to make appropriate and timely management decisions.

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