Case Report

Early Prenatal Diagnosis of Acrania by Radiologist in Peripheral District and Remote Areas

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How to cite this article: Ami Dave, Janardan Joshi, Tejas Kothawade, Purva Patel. Early prenatal diagnosis of acrania by radiologist in peripheral district and remote areas. International Journal of Contemporary Medicine Surgery and Radiology. 2022;7(4):D4-D5.

A B S T R A C T

Introduction: Acrania is a lethal congenital malformation characterized by partial or complete absence of flat bones in the cranial vault. In acrania, cerebral hemispheres develop completely and they are abnormal. Ultrasound allows early diagnosis of this anomaly.

Case Report: We report a 23 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.

Conclusion: Acrania is a rare lethal congenital anomaly characterised by an absence of the calvarium. Ultrasound allows early diagnosis of this anomaly. , an early identification is crucial for the patient counseling and making timely decision about the pregnancy.

Keywords: Acalvaria, Acrania, Anencephaly, Encephalocele, Meroacrania

INTRODUCTION

Acrania is a congenital malformation which is characterized by partial or complete absece of flat bones in the cranial vault where in the cerebral hemispheres develop completely but they are abnormal.Acrania occours as a result of an ectodermal and mesodermal development abbreation after neural tube closure. For the diagnosis of acrania there should be perfectly normal facial bones , a normal vertebral coloumn without fetal skull , and the volume of brain tissue should be equivalent to atleast one-third of gestational age matched brain size.Acrania is a lethal entity and the diagnosis can be made sonographically in the first trimester.

CASE REPORT

Hospital based antenatal ultrasonographic study was done in approximate 4400 patients during the period of January 2022 to December 2022 in the department of radiodiagnosis, Bhavsinhji civil hospital ,Porbandar, Gujarat.Amongst which a single case was reported with fetal Acrania .A 23 year old female was referred to our ultrasound department. Family history was non-corroborative. There was no history of intake of teratogenic drugs and other relevant past illness. and there was no significant family history of congenital anomalies. On ultrasound scan she was found to have an anencephalic fetus with a soft tissue mass resemble to brain tissue attached to the cranial end of the fetus. There was a single live fetus with a complete, well-formed brain including lateral ventricles, sulci, interhemispheric fissure, and brain convolutions. But there was no cranium and the brain was covered with a thin membrane. The brain had normal vascular pattern with the normal circle of Willis on color Doppler scan. Both orbits were equal in size, shape, and they were placed symmetrically. Fetal nose, nasolabial folds, and other facial structures were recognized and showed symmetry. The cervical, thoracic, lumbar spine, sacrum appeared normal, and had morphologically normal spinal canal. No obvious masses were seen attached to or arising from the spine. The long bones were appeared normal and femur length was compatible with the period of gestation. The four chamber view of the heart, liver, stomach, and bowels appeared normal and there were no ventral body wall defects. Genito-urinary structures appeared normal. Fetal kidneys were normal position, size, shape, and contour with normal pelvicalyceal system. There was well-distended bladder . No other gross abnormalities were detected. There was normal amount of liquor with amniotic fluid index of 13 cm. The placenta was in anterior location with normal thickness. Umbilical cord appeared normal and had good umbilical Doppler activity. Parents were counselled regarding the condition and the poor prognosis of the baby.

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. The fetal cranium is not fully calcified before 10–11 weeks; therefore, a first-trimester diagnosis must be made with caution. 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

ISSN (Online): 2565-4810; (Print): 2565-4802 | ICV 2019: 98.48 |

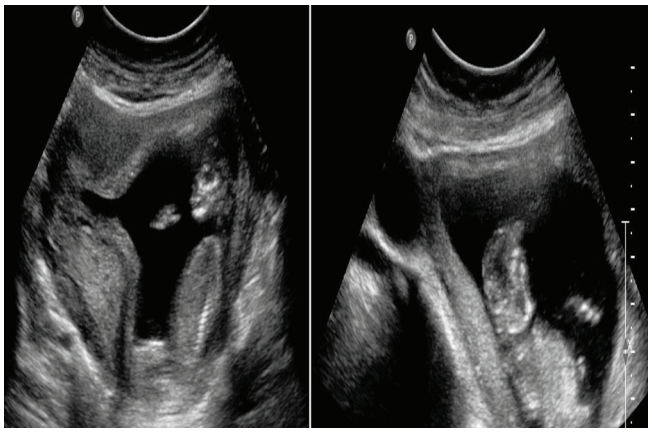

Figure 1 and 2: Ultrasound Images showing a fetus with Acrania

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 .

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 fluid. 3-D Ultrasound may contribute to early detection of fetal acrania and provide a novel visual depiction of this defect after reconstruction. 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 hypophosphatasis 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 diagnosis.

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 calcified. Antenatal identification allows the clinician to make appropriate and timely management decisions.

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Source of Support: Nil; **Conflict of Interest:** None

Submitted: 30-10-2022; **Accepted:** 28-11-2022; **Published online**: 30-12-2022