CASE REPORT

# FOETAL ACALVARIA: A CASE REPORT

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## Abstract

Foetal acalvaria is an extremely rare congenital abnormality characterized by the complete or partial absence of skull bones with complete but abnormal development of the brain. A 27 year old multiparous woman was found to have an anencephalic foetus at 12 weeks of gestation. Foetal ultrasound scan at 36 weeks revealed a well formed brain without a cranium. The baby was delivered by cesarean section. The brain was displaced posteriorly due to the absence of supporting skull bones and was covered by a thick membrane. The neonate expired few hours after delivery. Identification of acalvaria antenatally allows the clinician to plan an appropriate timely management.

## Introduction

Acalvaria is an extremely rare congenital malformation characterized by an absence of calvarial bones, dura mater and associated scalp muscles in the presence of basal skull bones, facial bones and complete but abnormally developed cranial contents covered by a scalp [(1)]. It occurs at a frequency of 1 in 100,000 births [(2)]. Since, acalvaria is usually a fatal anomaly and with only few survivors with this condition, treatment is limited. So, antenatal identification allows clinicians to plan an appropriate timely management.

## Case Presentation

A 27 year old non-diabetic, multiparous woman with a previous caesarian section at term for foetal distress was found to have an anencephalic foetus with a soft tissue mass at the cranial end of the foetus at 12 weeks of gestation. Patient was given routine antenatal care and at 36 weeks she had her normal obstetric examination and underwent foetal ultrasonography for further evaluation of the foetus.

A single live foetus was seen in cephalic presentation with longitudinal lie. Ultrasound scan revealed a complete, well-formed brain including brain convolutions, interhemispheric fissure, lateral ventricles

and the sulci **(Figure1)**. But there was no cranium and the brain was covered with a thick membrane. The brain showed normal vascular pattern with the circle of Willis on Doppler scan. Doppler waves were also normal. Facial structures appeared normal. Both orbits were symmetrically placed and equal in size and shape. No defects or masses were noticed in the foetal spine. Long bones appeared normal and the femur length was compatible with the period of gestation. The four chamber views of the heart, stomach, liver and bowels appeared normal and there were no features of ventral body wall defects. Genito-urinary structures appeared normal with a well distended bladder and male external genitalia. No other major abnormalities were detected.

There was normal amount of liquor and placenta was anterior with normal thickness. Umbilical cord appeared normal with good doppler activity. X-ray abdomen of the mother also revealed foetal acranium. The rest of the foetal skeleton was well formed with normal mineralization and there was a large soft tissue mass occupying the maternal true pelvis **(Figure 2)**.

Parents were counselled regarding the condition of the baby. A fully mature baby weighing 2.8 kg was delivered by an elective lower segment caesarian section **(Figure 3)**. Baby cried at birth and did not need initial resuscitation.

The baby's brain was displaced posteriorly due to the absence of supporting cranial bones. There were no frontal, temporal, occipital and the parietal bones in the foetal head. The brain was covered by a thick layer of skin. Facial structure and the rest of the body structures were completely normal. Baby had well-coordinated body movements. Baby's respiration was regular and there were no heart murmurs suggestive of cardiovascular involvement. Baby was admitted to the special care baby unit following delivery with a plan for conservative management. The baby developed sudden cardio respiratory arrest a few hours later and expired. A pathological postmortem was not done as parents did not consent.

## Discussion

There is no identified cause for foetal acalvaria[3]. It is a form of a congenital malformation that occurs as a result of faulty migration of the membranous neurocranium with normal placement of the embryonic ectoderm. This results in the formation of cranial contents in the absence of membranous bones but with an intact layer of skin over brain parenchyma [4].

Therefore, foetal acalvaria is considered to be a post neutralization defect [5]. The absent cranial bones may vary from case to case but, frontal, occipital and parietal bones are invariably absent [6].Acalvaria is sometimes confused with anencephaly, encephalocele and acrania[7]. In anencephaly, the cerebral tissues are completely absent while in encephalocele, the cranial vault is always detected and part of the brain protrudes outside. Acrania is the closest differential diagnosis and the two have been used interchangeably in medical literature many times [8].

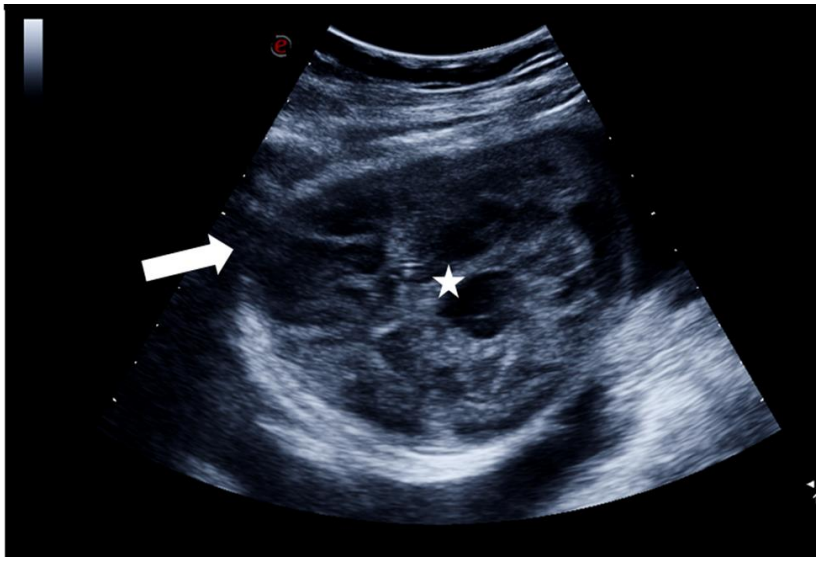

**Figure 1.** Foetal acalvaria; gray scale ultrasound of the foetal brain (3.5 mhz probe) in axial plane. Gestational age 38 weeks. No skull seen around the brain (arrow). Asterisk indicates the inter-hemispheric fissure

In acalvaria brain is covered by the scalp and in acrania brain tissues are exposed to the exterior.

Furthermore, acrania is essentially lethal and acalvaria could be consistent with life [9]. Acalvaria and acrania can be diagnosed as early as 12 weeks of gestation by ultrasound scan. Normal brain tissue seen on the coronal plane of the fetus results in the "Mickey Mouse" sign. The semicircular structures floating above the fetal face resemble the rounded ears of the Mickey Mouse [10].

Considering the prognosis, even though acalvaria is usually fatal, there have been few cases of survival. Some of these survivors were severely cognitively impaired and physically disabled.

Acalvaria has been reported in association with other abnormalities such as

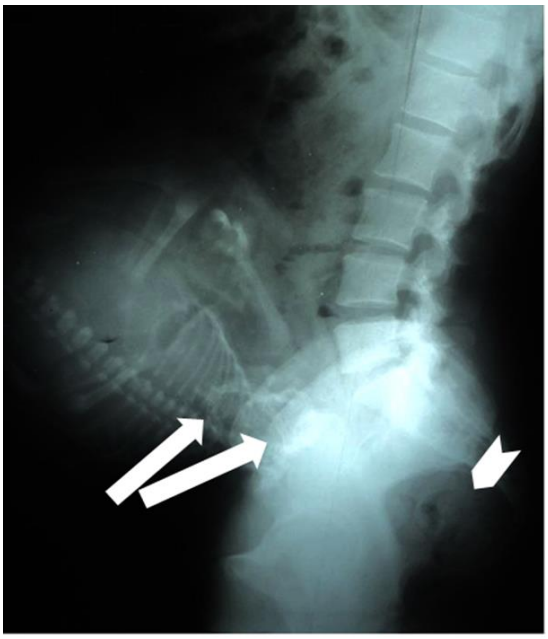

**Figure 2.** Foetal X Ray in lateral view. White arraows indicate the mandible and facial bones. Arrowhead indicates the foetal brain occupying the true pelvis of the mother.

micropolygyria, facial clefts and cardiac abnormalities, and hence need thorough clinical evaluation before prognosis is predicted.

**Figure 3.** Acalvaria; demonstrating the absence of cranium with developed brain covered with the scalp

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