Prenatal diagnosis of acalvaria

Abstract

Acalvaria, a rare and severe congenital anomaly, is characterized by the absence of cranial flat bones, muscles, and dura mater, while retaining a normal cranial base and facial bone development. Despite its rarity, the prenatal diagnosis of acalvaria is feasible, with potential implications for management. This article provides an overview of acalvaria, including its undefined etiology possibly linked to post-neurulation defects. Although surgical interventions have not been documented in the neonatal and early infancy stages, understanding and diagnosing acalvaria prenatally hold significance in comprehending its clinical course and prognosis.

Keywords: acalvaria, congenital malformation, prenatal diagnosis, etiology, postneurulation, prognosis

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Introduction

Acalvaria, an exceedingly uncommon congenital malformation, represents a perplexing anomaly characterized by the absence of cranial flat bones, dura mater, and muscles [1,2]. Its incidence is strikingly low, occurring in less than 1 out of every 100,000 births [1]. The rarity of acalvaria, coupled with its distinctive features, underscores the importance of early and accurate detection, particularly through prenatal diagnostic methods. The intricate nature of this condition beckons for further exploration into its etiology, clinical implications, and potential therapeutic strategies. As we delve into the depths of acalvaria, we aim to unravel its enigmatic origins and shed light on its clinical trajectory.

Case Presentation

Mrs. A.H, a 32-year-old woman with blood type O positive, presented without notable pathological history. She had a first-degree consanguineous marriage, and there was no mention of hereditary diseases in the family during the medical interview. She has had five pregnancies and three live births, with a history of uterine scarring. Her past medical history includes a pregnancy termination in 2011 at 15 weeks of gestation due to anencephaly/exencephaly. The current pregnancy was not under medical supervision, with no ongoing treatment or folic acid supplementation. At 13 weeks of gestation, she presented for a first-trimester ultrasound examination **(FIGURE 1)**.

Upon examination, the patient was conscious, hemodynamically stable, without bleeding, and reported no functional complaints. The ultrasound revealed a progressing mono-fetal pregnancy at 13 weeks, with a Crown-Rump Length (CRL) of 51.8 mm, nuchal translucency of 1.7 mm, femur length of 6.7 mm, and Bi-

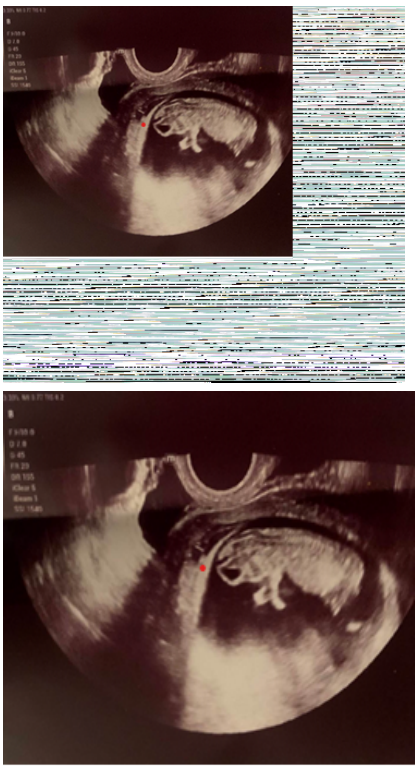

FIGURE 1.
Ultrasound images showing acalvaria.

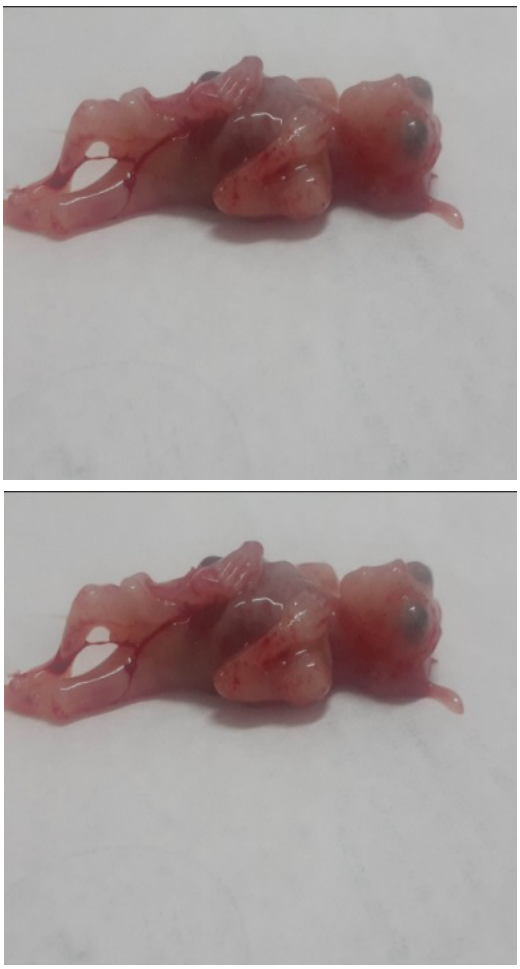

FIGURE 2: Embryo with acalvaria (Post Medical termination of pregnancy).

parietal Diameter (BPD) of 19.4 mm. The trophoblast appeared diffusely, and the amniotic fluid was of normal quantity. Morphological assessment indicated a partial to complete absence of the cranial vault, with a brain of normal diameter and varying absence of cranial bones.

After counseling the couple regarding the diagnosis of a lethal malformation, a therapeutic termination of pregnancy was deemed appropriate. The patient received cytotec and subsequently expelled a fetus along with the placenta. The placenta was sent for fetal pathology examination, and the results revealed no detectable chromosomal aberrations **(FIGURE 2)**.

Discussion

Acalvaria is a rare yet severe congenital malformation whose etiology remains elusive to this day. It is important to note that closure of the anterior neural pore occurs around the fourth week, followed by the migration of mesenchymal tissue beneath the underlying ectoderm, destined to form the future cerebral hemispheres. The ectoderm gives rise to the skin and scalp, while the mesenchyme contributes to muscle and bone formation. Anomalies of the neural tube are categorized into two subgroups: open defects due to neurulation abnormalities that occur prior to neural tube closure, which are more common and include anencephaly and myelomeningocele, and closed defects due to post-neurulation abnormalities, which are less frequent, including acalvaria. The latter theory is widely accepted, suggesting a defective migration of mesenchyme resulting in the absence of the calvarium. The embryonic ectoderm remains intact, along with the cranial base bones and facial bones. Other theories propose that it may result from primary non-closure of the neural tube. Acalvaria has also been associated with amniotic bands. No clear genetic factors have been established.

A gender predisposition towards females is noted in the literature. To date, the preventive role of folate acid in acalvaria remains unproven. Differential ultrasound diagnoses include anencephaly, encephalocele, osteogenesis imperfecta, and hypo-phosphatasia. The key differentiating ultrasound feature between acalvaria and anencephaly is the presence of intact cerebral hemispheres. Acalvaria displays a

"Mickey Mouse" appearance on ultrasound, with absence of flat cranial bones appearing as artifacts posterior to head structures, and the presence of cranial content covered by an echogenic layer representing the skin.

A novel prenatal diagnostic method involving Alpha-Fetoprotein (AFP) levels has been proposed. In normal pregnancies, AFP levels are low, but in acalvaria, they are elevated. This increase is likely due to the destruction of central nervous system tissues by amniotic fluid, though this method remains a topic of debate.

■ Treatment

To date, acalvaria has been described as a lethal anomaly. No surgical interventions to correct the cranial abnormality have been reported during the neonatal and early childhood periods. Surgical treatment, including bone grafting or plastic surgery, should be reserved for school-age children (4 years-5 years old). Treatment involves soft tissue augmentation coupled with skin grafting to facilitate dura mater protection by a layer of soft tissue, while maintaining a separate skin layer for future interventions. Subsequently, bone grafting is performed when the child reaches school age. The first surviving case was reported in Japan in 2004, where surgical closure of the scalp defect was performed along with shunt surgery for associated hydrocephalus. On follow-up, the child exhibited severe developmental delay, mental disorders, and growth retardation, last reported alive at the age of 11 [3,4].

■ Prognosis

Acalvaria is a fatal condition, with only a few survivors, often afflicted by severe neurological disorders.

Conclusion

Acalvaria represents a profound congenital anomaly, characterized by its rarity and potential diagnosis as early as the first trimester. To date, no preventive measures have been elucidated, and there exists no surgical intervention capable of curing this condition. The complex nature of acalvaria underscores the need for continued research to unravel its intricate pathogenesis and explore potential avenues for improved diagnostic and therapeutic approaches.

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