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Case Report

Fetal acalvaria with lateral cleft lip and palate: A rare presentation ☆

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ABSTRACT

We report a case of acalvaria diagnosed prenatally via ultrasound and MRI. Acalvaria is a rare, fatal congenital condition characterized by the absence of flat bones of the cranial vault, dura mater, and its associated muscles with an intact central nervous system. A 41-year-old gravida 5, para 2 + 2A, presented to us at 26 weeks gestation age (GA) with ultrasound findings of a fragile and hypomineralized skull in the fetus. The patient was not keen on whole-axon sequencing. Fetus magnetic resonance imaging (MRI) revealed large cutaneous/skull nonvisualization of the fetus skull, possibly acrania without anencephaly. She delivered via cesarean section at 37 weeks because of two previous cesarean sections. A female infant weighing 2650 g was born with an intact sac to minimize excessive external pressure to the brain tissue. A diagnosis of acalvaria with bilateral lateral cleft lip and palate was made postdelivery. The infant was managed conservatively per multidisciplinary discussion and expired 3 weeks later.

☆ Competing Interests: The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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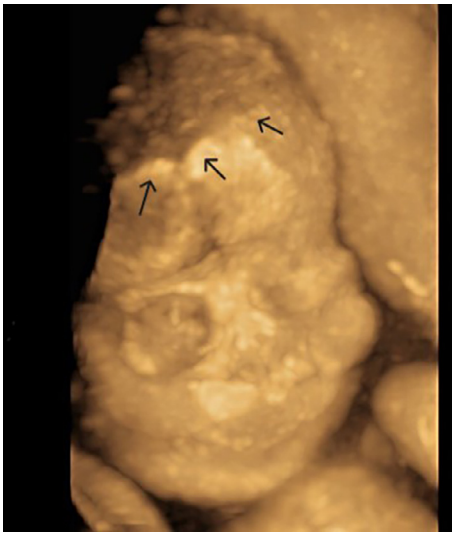

Fig. 1 – Absence of skull demonstrated on 3D ultrasound.

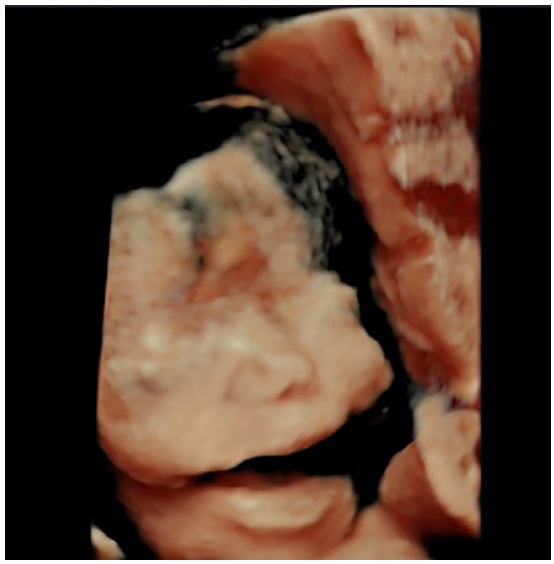

Fig. 2 – Bilateral transverse cleft lip and palate on 3D ultrasound.

Introduction

Acalvaria is a rare lethal condition characterized by flat bones of the cranial vault, dura mater, and associated muscles are absent, but the central nervous system is usually unaffected [1]. The disorder has unknown pathogenesis, is usually lethal, and is associated with other abnormalities. Thus, we herein present this uncommon fatal anomaly's clinical features so that undue associated morbidity can be avoided by prompt antenatal diagnosis and subsequent management.

Case report

A 41-year-old woman, gravida 5, para 2 + 2A at 26 weeks gestation, was referred to the fetal medicine unit for fetal microcephaly. Two-dimensional (2D) and 3D ultrasound examinations, using Voluson E6 (GE Healthcare), showed unusually well-defined brain structures in the hemisphere proximal to the transducer, suggesting a fragile hypomineralized skull. (Fig. 1) There's also a linear defect extending from both sides of the lip to the base of the ear, which is more apparent in 3D scans. (Fig. 2) Otherwise, there were no other features suggesting lethal skeletal dysplasia. The patient denied a family history of genetic disease or congenital anomalies. The patient was not keen on whole-axon sequencing prenatal diagnosis offered. A fetal magnetic resonance imaging (MRI) done at 28 weeks reported a significant cutaneous defect and nonvisualization of the fetus's skull at the temporoparietal part with bilateral lateral cleft lip and palate. (Fig. 3) The diagnosis of bilateral lateral cleft lip and palate with acrania without anencephaly was made, as a thin membrane was seen covering the fetal brain. Cesarean section was done at 37 weeks gestation as the patient presented in labor with 2 previous cesarean sections, delivering the baby's head in an intact sac to minimize excessive external pressure to brain tissue.

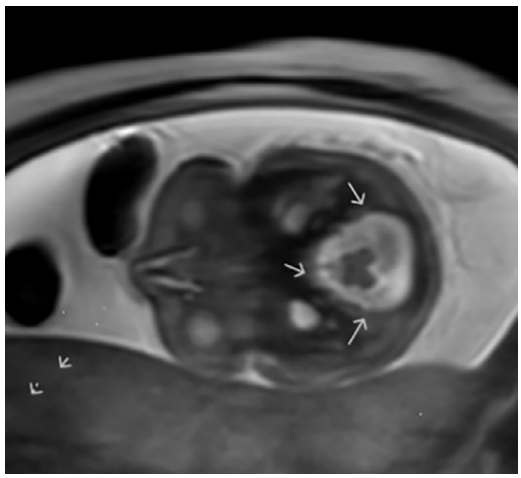

Fig. 3 – MRI image showing and nonvisualization of the fetus's skull at the temporoparietal part.

A female live infant weighing 2650 g was born with acalvaria (Fig. 4) and transverse cleft lip and palate. (Fig. 5) Based on a multidisciplinary discussion with the plastic surgeon, neurosurgeon, and Obstetric and Gynecology (O&G) teams, conservative management was opted for as the defect was nonoperable. The neonate was given supportive care and passed away 3 weeks later.

Discussion

Exencephaly, anencephaly, acrania, and acalvaria are conditions referred to as congenital cranial vault defects [1]. Acalvaria is a rare condition with less than 1 in 100,000 incidences [2]. It was sparingly reported in the literature, with

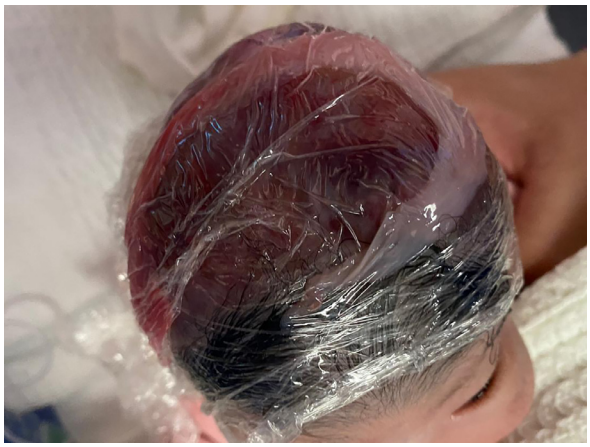

Fig. 4 – Acalvaria, covered with plastic to protect the exposed brain tissues.

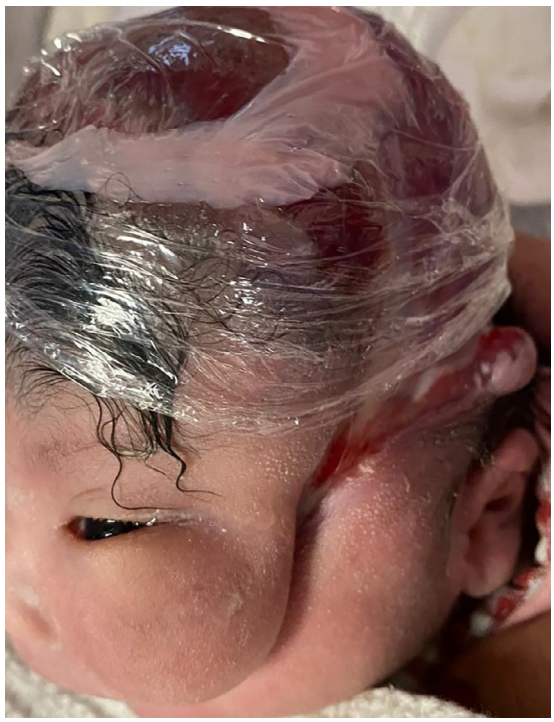

Fig. 5 – Transverse cleft lip up to the tragus of the ear.

only a handful of case reports [3]. The epidemiological survey suggests a female predilection. The pathogenesis of acalvaria is not precisely known [4]. Some investigators suggested the theory of postneurulation defect to explain the embryological basis of this malformation [5]. It is suggested that instead of migrating under the ectoderm, which forms skin and scalp, the mesenchymal tissue develops into skull bones and associated structures. Hence, this faulty mesenchyme migration in the presence of ectoderm leads to an absence of the calvarium but an intact layer of skin over the brain parenchyma [5]. Other theories suggest that it results from the primary nonclosure of the neural tube or maybe a part of a spectrum of anencephaly [6].

Acrania is a fatal anomaly since neurological abnormalities exist despite normal intracranial contents. So far, only a few live cases in the world have been reported in the literature, in which all of them are severely mentally retarded and disabled [7]. Unfortunately, this defect cannot be prevented by consumption of folic acid [5].

Prenatal diagnosis by ultrasound is challenging as the fetal brain shape and structures are intact. Hence, most cases reported in the literatures were diagnosed postdelivery. Diagnosis of acalvaria is only possible by second-trimester ultrasound after the mineralization of skull bones occurs [7]. It is usually associated with other congenital malformations such as cleft lip and palate, lung malformations, hydrocephalus, amniotic band syndrome, holoprosencephaly, and many more [4]. In this case, it is associated with bilateral lateral cleft lip and palate due to failure of the maxillary and mandibular processes of the first and second branchial arch fusion or disruption in the processes after fusion [8].

Diagnosis with ultrasound can be ambiguous, especially in early trimesters. Hence, simultaneous fetal MRI is increasingly utilized for confirmatory diagnosis when ultrasound results are undefined [1]. However, accurate diagnosis in utero is essential for an appropriate multidisciplinary team management plan and parental counseling. It is important to explain to the parents that acalvaria is usually sporadic and is not associated with chromosomal anomalies [1]. However, since acrania is a fatal condition, early recognition is crucial to aid parents in making informed decisions regarding the continuation of pregnancy.

Conclusion

We present a rare case of an infant diagnosed prenatally with acalvaria and transverse cleft lip and palate who lived until 3 weeks on conservative management. This case illustrates the hardship in making an exact diagnosis prenatally with ultrasound alone, but it is made possible with the help of MRI. Detecting fetal anomalies in utero is to provide appropriate counseling to the parents.

Patient consent

Written consent has been obtained from the patient's parents, and approval to publish was obtained from the Ethical Committee, Ministry of Health, Malaysia and following local legislation.

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