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Case Report

A case report on fetal acrania

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ABSTRACT

Fetal acrania is a rare but lethal congenital malformation within the acrania-exencephaly-anencephaly spectrum, characterized by complete absence of the cranial vault, meninges, and overlying scalp. The reported incidence is 1.4-2 per 1000 births in India, making it one of the most common neural tube defects after cardiac anomalies. Early detection through ultrasonography is crucial for counselling, prevention of maternal complications, and guiding reproductive planning. A 21-year-old primigravida presented at 16 weeks of gestation for routine antenatal care. She was unbooked case. Diagnosis of acrania was established by ultrasound, and the patient was managed with medical termination of pregnancy following counselling. Early sonographic diagnosis is vital for appropriate counselling and decision-making. Pre-conceptional folic acid supplementation remains the most effective preventive measure against neural tube defects.

Keywords: Acrania, Neural tube defects, Preconceptional counselling, Prenatal ultrasonography, High-risk pregnancy, Folic acid supplementation

INTRODUCTION

Foetal acrania is a fatal congenital anomaly characterized by complete or partial absence of the cranial vault, meninges, and overlying scalp, exposing the developing brain to amniotic fluid. It forms part of the acrania-exencephaly-anencephaly spectrum, a continuum that begins with absence of calvaria bones, progresses to exencephaly, and culminates in anencephaly due to cerebral degeneration.[1]

The global incidence of neural tube defects ranges from 1-5 per 1000 live births, with variation by region and maternal risk factors.[2]

In India, the incidence of acrania has been reported between 1.4 and 2 per 1000 births, ranking second only to cardiac anomalies among congenital malformations.[3]

Risk factors include maternal folate deficiency, obesity, diabetes mellitus, hyperthermia, anticonvulsant exposure, and low socioeconomic status. Recurrence risk is estimated at 3-5% in subsequent pregnancies without folic acid supplementation.[4]

The underlying pathophysiology involves failure of mesenchymal migration and differentiation in early embryogenesis, resulting in absent membranous bones of the cranial vault.[5]

Prenatal ultrasonography is gold standard for diagnosis. Acrania can be identified as early as 8-10 weeks by the absence of ossified calvarium, frog-eye appearance from prominent orbits, and lack of echogenic cranial margins.[6]

Differentiation from skeletal dysplasia such as osteogenesis imperfecta and hypophosphatasia requires systematic evaluation of ossification centers and cerebral structures in line with ISUOG guidelines.[7]

Given its fatal prognosis, counselling is essential to inform parents about the nature of the anomaly, the option of

termination when diagnosed early, and the importance of preconceptional folic acid supplementation, which reduces recurrence of neural tube defects by up to 72%.[8]

We present here a case of foetal acrania diagnosed in the second trimester, underscoring the importance of antenatal screening and preventive measures.

CASE REPORT

A 21-year-old primigravida presented at 16 weeks of gestation for routine antenatal care. She was unbooked, with no previous antenatal visits, no dating scan and no history of periconceptional folic acid supplementation. There was no past medical or surgical illness. On general examination, pallor was noted. Her BP-110/70 mmHg, PR-84 bpm Her BMI was 20.6 kg/m^2. On Systemic examination CVS-normal, RS-normal and CNS-no abnormality detected. Per abdomen, the uterus corresponded to 16–18 weeks of gestation and was relaxed. Foetal heart rate was recorded at 136 beats per minute.

Laboratory investigations revealed haemoglobin 8.1 g/dL, total leukocyte count 11,800/cumm, and platelet count 4.21 lakhs/cumm. Blood group was A positive. Urine routine examination was normal. Random blood sugar was 101 mg/dL. Viral serology for HIV, HBsAg and VDRL was non-reactive. Thyroid-stimulating hormone was 1.26 µIU/ml. Coagulation profile was within normal limits (PT 14.6 s, aPTT 30.0 s, INR 1.0).

Ultrasonography showed a single live intrauterine foetus of 15 weeks 6 days with absence of cranial vault bones, consistent with foetal acrania. Placenta was anterior, grade I, and foetal heart rate was 111 bpm. Differential diagnoses such as osteogenesis imperfecta and congenital hypophosphatasia were excluded by the presence of preserved brain parenchyma and typical cranial features (Figure 1 and 2).

Management

The patient and her family were counselled regarding the poor prognosis. Informed consent for termination of pregnancy was obtained. She was admitted and transfused one unit of packed red blood cells in view of anaemia. Medical termination was initiated with oral mifepristone 200 mg, followed by induction with Foley's catheter and intravenous oxytocin infusion.

She expelled a foetus weighing 400 g (Figure 3 and 4). Gross examination revealed absence of the cranial vault and frog-eye appearance. Infantogram confirmed absence of cranial bones, consistent with acrania (Figure 5). The postpartum course was uneventful. Before discharge, the patient was counselled regarding preconceptional folic acid supplementation and the importance of early antenatal care in subsequent pregnancies.

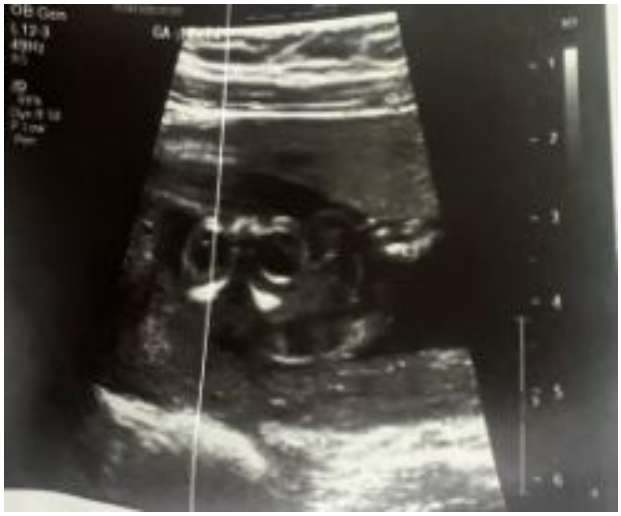

Figure 1: USG of frog eye appearance.

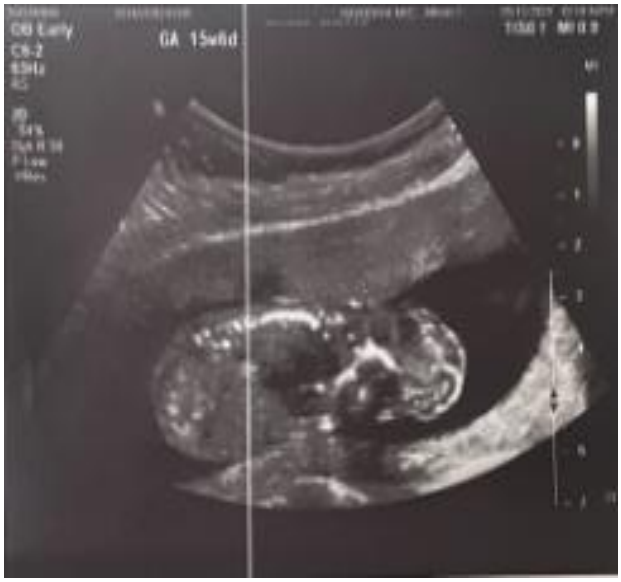

Figure 2: Absence of cranial vault bones.

Figure 3: Fetus with absent cranial bone.

Figure 4: Foetus with acrania.

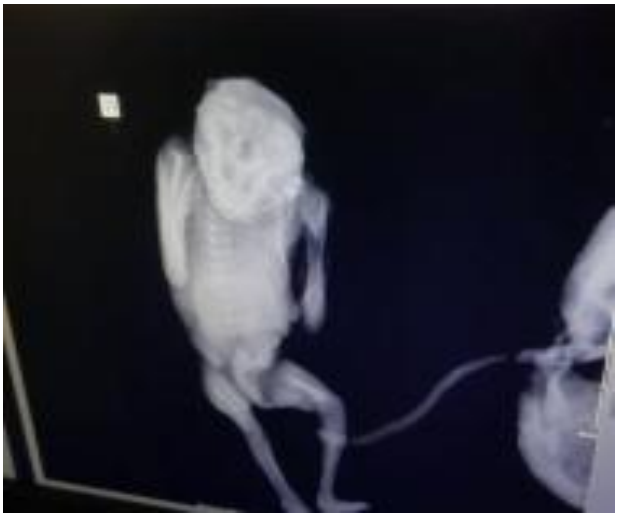

Figure 5: Infantogram of the foetus.

DISCUSSION

Acrania is the most common anomaly within the acrania-exencephaly-anencephaly spectrum (AEAS). This spectrum represents a continuum of lesions beginning with the absence of the cranial vault (acrania), progressing to exposure of the virtually normally developed brain into the amniotic cavity (exencephaly), and culminating in anencephaly as a consequence of progressive brain destruction. In foetal acrania, there is a partial or complete absence of the flat bones of the cranial vault. Even if cerebral tissue develops, it is structurally abnormal. The cerebral hemispheres fail to form properly, although the brainstem, cerebellum, and cranial nerves are usually preserved. The condition is fatal, and if pregnancy continues, acrania inevitably progresses along this spectrum to anencephaly.[9]

Several studies have demonstrated the ability of first-trimester ultrasonography to identify acrania as early as 9–10 weeks of gestation. Monteagudo and Timor-Tritsch reported the classic "Mickey Mouse" or "frog-eye" sonographic sign as a reliable marker for early diagnosis. Differentiation from skeletal dysplasia is important, as conditions such as osteogenesis imperfecta also present with poor ossification of the calvarium. In osteogenesis imperfecta, however, ultrasonography typically shows diffuse skeletal hypo mineralization, multiple long-bone fractures, shortened and bowed limbs, and a compressible calvarium, whereas in acrania the abnormality is restricted to the absence of cranial vault bones with preserved limb ossification.[10]

The psychological and ethical aspects of acrania diagnosis are equally important. Literature emphasizes that parents often face significant emotional distress upon receiving the diagnosis of a lethal anomaly. Counselling should therefore be conducted with sensitivity, providing clear information about prognosis, the option of medical termination, and preventive strategies for future pregnancies. Genetic counselling also plays an important role, particularly in women with prior affected pregnancies, to discuss recurrence risks as well as the protective role of the periconceptional folic acid supplementations.[11]

In our case, the patient presented in the second trimester with moderate anaemia and was diagnosed with foetal acrania on ultrasound. Management involved correction of anaemia, counselling regarding the prognosis, and termination of pregnancy after informed consent. The diagnosis was supported by characteristic gross findings and infantogram following expulsion. The case underscores two critical issues: the importance of early antenatal registration and anomaly screening, and the role of pre-conceptional folic acid in reducing the burden of neural tube defects.[12]

CONCLUSION

Congenital anomalies can be reliably identified by prenatal ultrasonography. Early diagnosis is essential to provide appropriate counselling, prevent maternal morbidity, and assist families in making informed reproductive decisions. Daily intake of 400 micrograms of folic acid in the preconceptional period and continued through the first three months of pregnancy has been shown to significantly reduce the risk of neural tube defects, with preconceptional supplementation lowering recurrence risk by up to 72%. Regular antenatal care, including a nuchal translucency scan between 11-13^{+6} weeks and an anomaly scan between 18-22 weeks, plays a vital role in detecting congenital anomalies at an early stage. Early recognition enables timely counselling regarding the option of termination in affected pregnancies, thereby minimizing maternal complications and neonatal morbidity.

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