Acrania Without Anencephaly

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Abstract

Acrania is an extremely rare lethal developmental anomaly characterized by a partial or complete absence of calvarium with complete but abnormal development of brain tissue. Although acrania associated with anencephaly is a well recognized entity, isolated acrania without anencephaly is an extremely rare anomaly and its true incidence is unknown with only a few reported cases. We also discuss the difference between acrania and acalvaria with which it is has been often confused even in published literature.

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INTRODUCTION

Acrania (syn. exencephaly)is a developmental anomaly characterized by complete or partial absence of the neurocranium with complete but abnormal development of brain tissue. This rare lethal anomaly has important implications for obstetrical management and counseling. Prenatal ultrasound readily allows early diagnosis of this condition by the alert sonologist.

CASE REPORT

A 24 year old non-diabetic, non-alcoholic primigravida with normal obstetric examination came for fetal sonography. A single live fetus was seen with cephalic presentation .Liquor was adequate with closed internal os. Movements were sluggish and scanty. Femur length, abdominal circumference and head circumference all were matching with menstrually calculated gestational age of about 26 weeks. Ultrasound showed well formed cerebral hemispheres with lateral ventricles but without the echogenic rim of skull (Figure 1 & Figure 2).

Figure 1

Figure 1: show absence of the echogenic calvarium on sonography

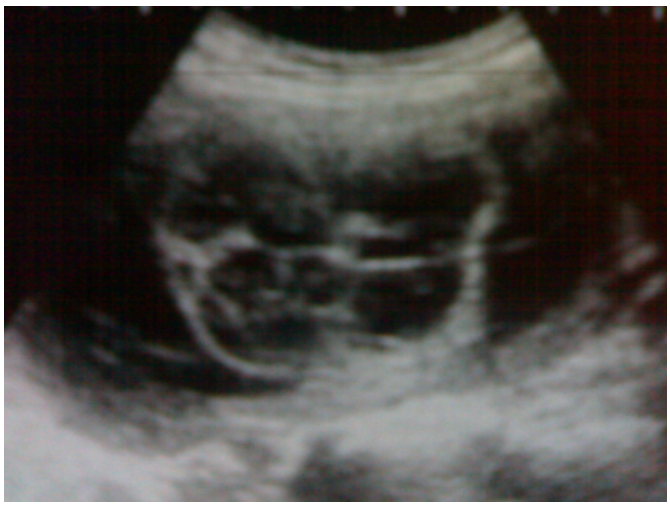
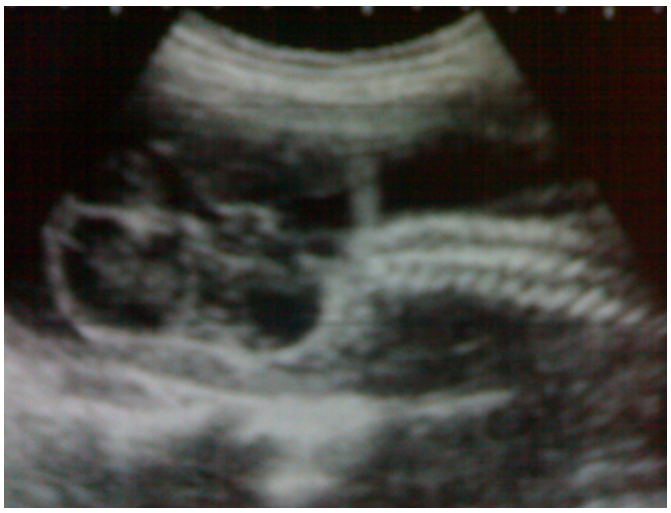


Figure 2

Figure 2: shows lack of the calvarium contrasted against the normal ossified spine on sonogram.

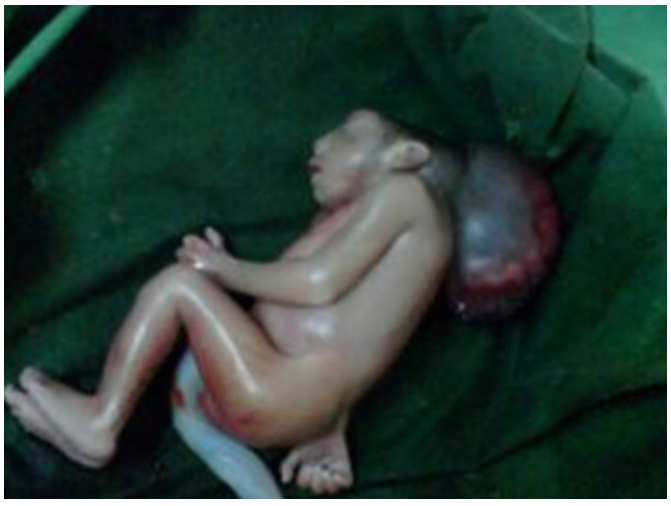


No other obvious internal anomaly was detectable. Fetal echocardiogram showed normal cardiac chambers. Fetal umbilical vessels showed normal flow on doppler.

A diagnosis of acrania was made and the fetus was immediately evacuated by vaginal route using prostaglandins. It was a female fetus with stillbirth. Skull vault and scalp was completely absent with pia covered brain tissue exposed to the exterior (Figure 3).

Figure 3

Figure 3: shows absence of the neurocranium with well developed brain covered with pia-arachnoid; the chondrocranium(base of brain) is well developed.



The brain has slipped posteriorly due to absence of supporting bones. Facies, body and limbs were externally normal with no other deformity. A postmortem internal examination could not be performed as the parents did not consent.

DISCUSSION

Although acrania associated with anencephaly is a well recognized entity with an incidence of about 1:10,000 births , isolated acrania is a rare anomaly and its exact incidence is unknown($_1$).Only few cases have been reported in world literature.

The neurocranium is composed of chondrocranium which forms the bones of the base and the membranous flat bones which encase the brain. Acrania affects the membranous flat bones and it is not uncommon to see rudimentary bones at the base of skull. Acrania occurs during the beginning of the fourth week of development when the anterior neuropore closes. The normal migration of mesenchymal tissue under the calvarial ectoderm(which itself remains membranous instead of forming the epidermis) does not occur. Thus calvarial bones of the skull ,related musculature and dura mater are absent. In the absence of induction from the neurocranium the cerebral tissue fails to differentiate into two hemispheres. The cerebellum, brainstem and the cranial nerves are normal , but the diencephalon and outer globe is abnormally small. A few cases of acrania has also been reported due to amniotic bands($_2$).

Prenatal diagnosis by US can be made by as early as 12 weeks by transvaginal sonography ($_3$) albeit exact differentiation from anencephaly can be confidentially made by 15-16 weeks only. The condition is diagnosed by absence of the calvarium which is seen as a high echogenic bright rim surrounding the brain tissue normally. In acrania a thin membrane is seen surrounding the cerebal hemispheres ($_4$). The demonstration of abundant, relatively well differentiated (though not completely normal)brain tissue helps differentiate acrania from the much more commoner anencephaly.

Multiple associated anomalies have been seen with acrania inconsistently like cardiac anomalies, club foot, cleft palate, tethered cord, ompahlocele and pentalogy of Cantrell(2).

The closest differential diagnosis includes anencephaly or a large cephalocele. In anencephaly cerebral tissue is completely absent while in cephaloceles cranial vault can always be detected and a part of brain is intracranial ($_5$). Distinction also has to be made from conditions characterized by lack of skull mineralization like hypophosphatasia or osteogenesis imperfecta type 2. In these skeletal dysplasias brain anatomy is normal and brain is

surrounded by a thin rim of soft tissue representing unossified skull and scalp. Bowing, fractures and shortening of long bones are usually present ([5]).

Although acrania and acalvaria have been many times used synonymously in medical literature, the two differ in the sense that in acalvaria scalp is intact and normal while in acrania (exencephaly) brain surface is exposed ([6]). Also, acalvaria could be consistent with life while acrania is essentially lethal. Some consider acrania to be a precursor to anencephaly due to chemical damage to the exposed brain while in acalvaria the brain is protected by the intact scalp ([7]).

Alpha fetoprotein levels are elevated in maternal serum, while urinary estriol is undetectable. Increased incidence in subsequent pregnancies has not been reported.

Acrania is uniformly lethal. Although two surviving cases have been reported these appear to be cases of acalvaria (rather than acrania) as the scalp was normal and intact in both these cases ([8]). Antenatal identification allows the clinician to make appropriate timely management decisions.

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