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Bone Defect of the Cranial Vault: Difficulty of the Diagnostic about a Case, and Revew of Literature

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Abstract

The bone defects of the cranial vault encompassed rare malformations including acalvaria, hypocalvaria, acrania, hypocrania, anencephaly and exencephaly. They are also described in some pathological entities such as aplasia cutis congenita of the scalp. We report an unusual case of cephalic malformation which combine defects of the skin, the dura mater, and the bones of the vault, with a malformation of the central nervous system. This unique case emphasizes a problem of nosological definition between the terms mentioned above. acalvaria, the acrania, the hypocalvaria and the aplasia cutis congenita. Thus, herein, we proceed to a literature review of bone defects of the skull and their differential diagnosis.

Keywords

Cranial Vault Defects, Acalvaria, Hypocalvaria, Aplasia Cutis Congenita, Congenital Malformation, Diagnosis

1. Introduction

The cranial vault bone defects encompass a group of rare congenital malformations where all or part of the cranial vault bones are absent. Depending on the extent of missing cranial vault bones, the corresponding nosological terms can be *acalvaria*, *hypocalvaria*, *acrania* or *hypocrania*. This can be associated with or without anencephaly and exencephaly. Another term found in the literature is aplasia cutis congenita of the scalp. Hence, its definitions remain controversial.

The differential diagnostic criteria bear on the presence or absence of abnormal skin, muscular, meningeal, and nervous tissues.

A calvaria is a rare congenital disorder characterized by the lack of development of the flat bones of the skull, that is membranous neurocranium, dura mater, and associated muscles in the presence of normal cranial contents and facial bones [1] [2] [3] [4]. Hypocalvaria refers more to hypoplasia than an absence of cranial bones [5]. Khadilkar [3] evoke an incomplete absence of the vault. Morever, in acrania, the bones of the vault are partially or entirely absent with a complete but abnormal development of the cerebral hemispheres [1]. The common location of the aplasia cutis congenita is on the scalp. It typically consist of the lack of skin on the scalp and the underlining tissues namely dura mater and skull bones [6].

We report an unusual case of cephalic malformation with skull bone and skin defects, in which we encountered difficulties in diagnosis and nosological definition. Hence, we perform a literature review on skull bone defects and aplasia cutis congenita of the scalp.

2. Patient Observation

Our case was a 2690-gram female newborn by caesarean at 37 weeks of gestation. She came to us at five hours of life with a diagnosis suspicion of occipital encephalocele. Her mother was 29 years old with no history of malformations on her previous pregnancies. There were only 2 prenatal medical visits with the first visit performed at 7th month of gestation. At this time, an ultrasound at 27 weeks of gestation suspected a hydrocephalus. No parent's consanguinity was found. Of note, her mother unsuccessfully attempted abortion using unspecified traditional medicines. On the physical examination, the newborn had a normal head circumference measuring 32 cm. Apgar was 7 and 8 at 1 minute and 5 minutes respectively. The patient had an8 cm diameter midline occipital skin defect associated with a defect of dura matter, an exposed brain parenchyma and cerebro-spinal fluid (CSF) leakage (**Figure 1**). The palpation of the surrounding skin found an absence of the underlying occipital and parietal bones. Additionally, the newborn had an adult-like facial feature. The rest of examination was normal. The brain CT scan revealed a major hypoplasia of the temporal, frontal and occipital bones, and a minor hypoplasia of the temporal bones. There were no anomalies of skull base and facial bones (**Figure 2**). Moreover, the newborn presented brain malformations such as an agenesis of the corpus callosum and a ventricular ectasia .Under general anesthesia, we performed a surgical repair of the skin with rotational flap at day 4 of life. The immediate post operative outcome was uneventful. The scar tissue was good after 10 days (**Figure 3**) and the patient was discharged with closed follow-ups. However, at 1 month of life, the patient developed a hydrocephalus treated successfully by ventriculo-peritoneal shunt. The follow-up 4 months later was uneventful. At 12 months of life, the patient developed a community acquired pneumonia and died.

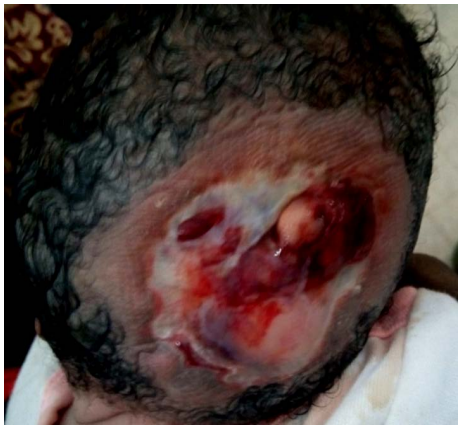

Figure 1. Occipitotemporal skin defect about 8 cm in diameter with bone and dura defect, putting the brain exposed (image obtained with the parent's informed written consent).

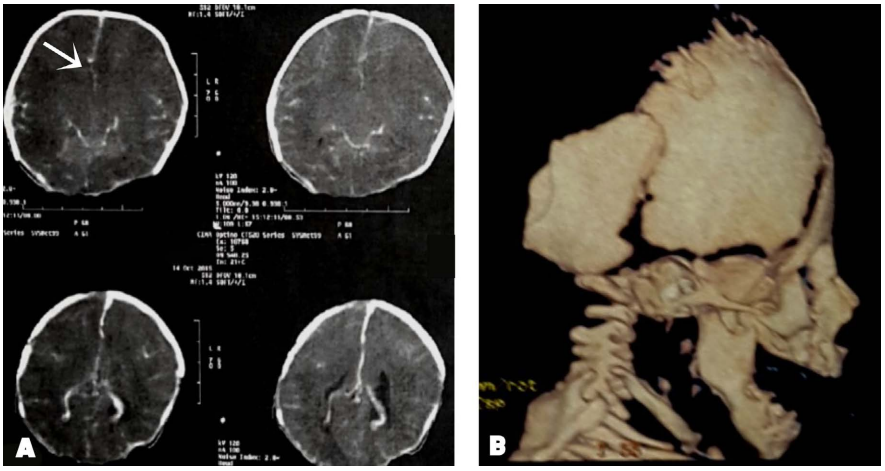

Figure 2. Brain CT scan showing parenchymal and bony malformations (A) Agenesis of the corpus callosum and diencephalic structures (showed with the white arrow) (B) Cranioencephalic CT scan; 3D reconstruction. Hypoplasia of the bones in the vault (occipital, parietal, temporal, frontal).

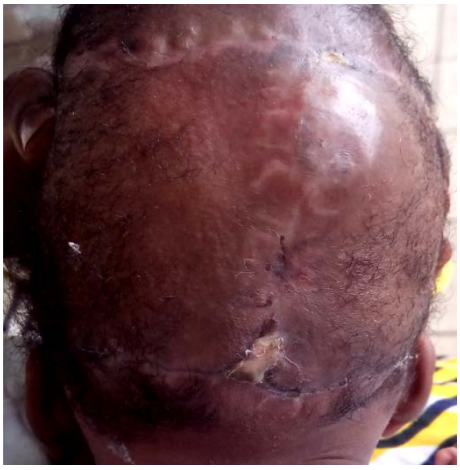

Figure 3. Photograph of the posterior part of the head, showing the closure of the skin defect (image obtained with the parent's informed written consent).

3. Discussion

This reported case exhibits a problem of nosological definition. It combines central nervous system anomaly, skin and dura matter defects with vault bones defects. The terms acalvaria, hypocalvaria, acrania or aplasia cutis congenita of scalp refer to cephalic malformations in which vault bones defect are involved. Our supposed diagnosis was first aplasia cutis congenital and secondly acalvaria or acrania. Crowder [7] noted these nosological and diagnostic difficulties. Thus the final diagnosis in his reported case was made after autopsy.

The diagnosis criteria of acalvaria include the absence of the vault bone, a normal chondrocranium development and the presence of both cerebral hemispheres. Hence, in Acalvaria, the facial bones are present, and the intracranial content is normal [1] [2] [4] [8] [9]. However, some brain morphological abnormalities may rarely be associated. Thus, holoprosencephaly, micropolygiria, hydrocephalus has been reported [2] [3]. In our case, an agenesis of the corpus callosum existed; hydrocephalus was diagnosed after the wound closure like it is described in neural tube closure anomalies. Yet, the exact pathogeny is still obscure. Several theories are suggested. Among them, is the theory of post neurulation defect (after day 24 - 26 of gestation).In normal condition, after the closure of the anterior neuropore (occurring within the week 4 of gestation), the mesenchymal tissue migrates under the ectoderm (which forms the skin and the scalp); but, instead, the mesenchymal tissue develops between the cranial bones and the surrounding muscles. That abnormal migration leads to the absence of elaboration of flat cranial bones and surrounding muscles [2] [3] [10].

So, as the scalp is present in Acalvaria, we cannot conclude in that diagnosis in our case. But Hawasli [11] reported a case of acalvaria associated with skin defect which he also named "acrania". Some cases of Acalvaria with skin defect were reported associated with amniotic band syndrome and Angiotensin Converting Enzyme (ACE) intake during the pregnancy [5] [8] [12]. For that reason, Moore [10] made a distinction between primary acalvaria (due to abnormal migration of the embryonic mesoderm) and secondary "Acalvaria" (due to cerebral hypoxia that leads to the anomaly of the neural ridge). Acalvaria has a bad prognosis; it is a source of spontaneous abortion and is considered as a fatal disease in newborns [13]. Hence, several cases are reported in foetus. So far, some survival cases of Acalvaria have been reported [12]. An early diagnostic can be performed since the week 12 of gestation by a transvaginal echography showing normal cerebral hemispheres [2] [3] [12]. In Acalvaria, Alpha Foeto-Protein rate is high and Free Oestrogen is not detectable. Those findings were not observed in our case. According to Gupta [13], weshould look for history of X ray exposition in mothers. It was not noticed in our case.

Acrania is defined as the partial or entire absence of the vault associated with an abnormal development of the chondrocranium, cerebral hemispheres are present but with brain anomalies [1] [9]. An early diagnosis can equally be achieved by a transvaginal echography which shows structural brain anomalies

[8]. So, in fetus, the following criteria can define acrania: a normal face perfectly bone, a normal cervical column purpose without the fetal skull and a volume of brain tissue equivalent to at least one third of normal the brain size. Acrania is frequently associated with anencephaly and is not viable. Then, Acrania and Anencephaly are often confounded. In our case there is an abnormal development of the chondrocranium, the skull base was hypoplastic whereas both hemispheres are presents.

In Anencephaly, the skin and the skull bones are partially or completely absent and the nervous tissue is exposed [10]. Thus, Anencephaly cases features Acrania. Some authors state that Acrania could precede Anencephaly in promoting the mechanical and chemical destruction of the cerebral parenchyma by the amniotic fluid [1] [14] [15]. On another hand, some scientists assert that Anencephaly is a consequence of a failure of neural tube closure [2] [3]. But the folate insufficiency is not proved to be considered as a risk factor although Bianca [1] recommended a supplementation in folic acid before conception. Similarly, genetic factors haven't been incriminated in the etiopathogenesis of Acrania. In our case, the skin closure leads to an acute hydrocephalus as what we observed in the anomalies of the neural tube closure where those factors are observed. Acrania, as well as Acalvaria can be linked to a amniotic band syndrome [16], not founded in our reported case. The mechanism of Acrania remains controversial. Mannes [17] supposed that failed migration of the mesenchyme under the calvarian ectoderm during the 4th week of development would account for agenesis of the calvarian bone, the musculature, and the dura mater. On the other hand, Kurata [4], suggested that Acrania was due to a failure in mesenchyme differentiation overlying the brain later than the 5th week of gestation.

Anencephaly is closer to Exencephaly, in which there is an Acrania associated with a cerebral anomaly due to an anomaly of the skull base development [1] [9].

Hypocalvaria is defined as a partial lack of the vault bones [2] [5]. This term was suggested by Barr [5] in 1991 to name the cases of vault hypoplastic membranous bones independently of the state of the skull base and brain, described in some fetopathies linked to the intake of ACE (Angiotensin Converting Enzyme). The fetopathies has several manifestations. Etiological and pathogeny factors are various but although the pathogenic factors are still unknown; it may be a hypoxic process. This theory resembles the pathogeny of the secondary Acalvaria described by Moore [10]. Hypocalvaria could be more frequent than Acalvaria (of a better prognosis) [5] [9]; and seems to be adequate to describe our case report. But Burkhead [18] described a similar case he named "Aplasia cutis congenita" of the scalp.

Aplasia cutis congenita of the scalp is equally a rare disease. The scalp is described as the most frequent location of this disease. It is a focal lack of the epidermal tissue, dermal tissue, subcutaneous cellular tissue, vault bone (in 15% - 20% of cases) and the dura matter [18] [19] [20]. Other locations of the aplasia cutis congenita are the trunk and the limbs. The underlying mechanism is still

elusive. Some etiological factors have been reportedparticularly genetic, obstetrical, traumatic, vascular, and infectious factors, as other promoting factors such as drug intakes or noxious substances for the embryo [19] [21] [22]. Frieden [23] in 1986 classified cases of aplasia cutis congenita in 09 groups, our case gathers the criteria of the group 1.

Regardless of vault bone defects, the diagnostic can be done early by the obstetrical echography in the second semester of pregnancy when the mineralization of the cranial bones ended [8]. The problem remains the treatment of the bone defect. Even untreated, the evolution is generally without complications and simple wound care is suitable. Complications may arise and aggravate the prognosis such as hemorrhage, local infection, meningitis or more severe sagittal sinus thrombosis [21]. In aplasia cutis congenita a spontaneous development of bones has been described. So, the treatment is commonly medical, although in cases of large defects the therapeutic choice may be a dilemma [12] [22]. Meanwhile, for other cases, a surgical treatment may be required. The surgical treatment encompasses various methods such as bone graft reconstruction method, cranioplasty, use of bony growth stimulating agents. All those methods allow a spontaneous bony growth that shrinks the defect before the cranioplasty [15]. There is no concensus about treatment.

4. Conclusion

Congenital vault bone defects are quite rare. A review of the literature reveals several distinct appellations for those congenital diseases. As highlighted in our case, there is a need to clarify the nomenclature of those entities. The reported cases of survivors underline the concern for the therapeutic methods appropriate to the bone defects.

Ethic

This study has been approved by the local Institutional Review Board. The images of the patient were obtained with the parent's permission.

Conflicts of Interest

The authors declare no conflicts of interest concerning this study or the findings specified in this paper.

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