



Case Report

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Cranium bifidum occultum with severe hypoplasia of parietal bones associated to corpus callosum agenesis and seizures

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Abstract

Cranioschisis, or cranium bifidum is an unusual lesion. In this report the authors present a case of cranium bifidum occultum associated with corpus callosum agenesis and seizure, diagnosed in a newborn boy who had large bilateral unossified parietal bones defect. We present the brain CT and MRI imaging findings showing the severe hypoplasia of both parietal bones and the agenesis of corpus callosum. The cranial defect persisted for a year during the follow-up period with no significant change. The boy now has stable condition.

Keywords: Cranium bifidum occultum, Hypoplasia of parietal bones, Developmental skull defect, Corpus callosum agenesis, Pediatric radiology.

INTRODUCTION

Defects of calvarial intramembranous ossification are recognized as cranium bifidum and foramina parietalia permagna, and are due to mutations in the ALX4 and MSX2 genes^[1]. The term cranium bifidum literally means “cleft skull” and is used to designate a defective closure of the skull by analogy with that of its spinal counterpart (spina bifida), while cranium bifidum occultum consist in a rare entity in which there is a delayed ossification of parietal bones resulting in a confluent, midline cranial vault defect with an intact scalp, pericranium and dura^[2-4]. It has an incidence of 1-3 per 10,000 births^[5,6]. The aim of this work is to present the clinical and imaging findings of an interesting case of cranium bifidum occultum with severe hypoplasia of parietal bones, agenesis of corpus callosum and seizures.

CASE REPORT

A 34-days-old male born at 37 weeks' gestation following caesarean section for fetal distress, was admitted at our hospital with seizure and a large midline palpable skull bone defect (Fig 1) with no herniation of cranial contents.

The healthy mother reported an uncomplicated pregnancy with no exposures to drugs or alcohol during gestation.

On clinical examination the skull was soft with very large fontanelles, giving the impression of a nearly absent calvarium. The gyri and sulci of the cortex were easily palpated through the scalp. The findings of the remainder of the physical examination were normal.

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A plain skull radiograph showed a big midline bone defect in the fronto-parietal region due to the lack of parietal bones ossification.

An ultrasound scan of the brain revealed agenesis of the corpus callosum.

Computed tomography (CT) demonstrated severe hypoplasia of parietal bones showing a large defect situated at the vertex of the skull (Fig 2), as well as two defects of frontal middle line, one of them, lower and well circumscribed, measuring 9.5 mm in maximal dimension, with non-sclerotic margins and with no herniation of cranial contents through the skull defects (Fig3). Follow up clinical examination at one year demonstrated some decrease in the size of the lesion with no new associated findings. By 13 months of age, a repeat CT scan showed minimal new bone formation in the parietal area. 3D CT reconstruction of the skull display further delineated the bone defect (Figs 2, 3). CT also documented lack of sutural formation of contiguous portions of left parietal and occipital bones. CT also made suspect corpus callosum agenesis that was confirmed by MRI (Fig 4).

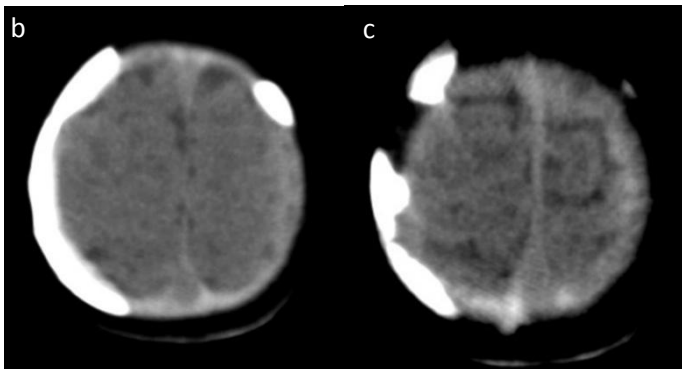


Figure 1: a- CT scout. Large lucency in the vertex region of the skull vault. b & c- Axial views 13 months later showing minimal bone growth.

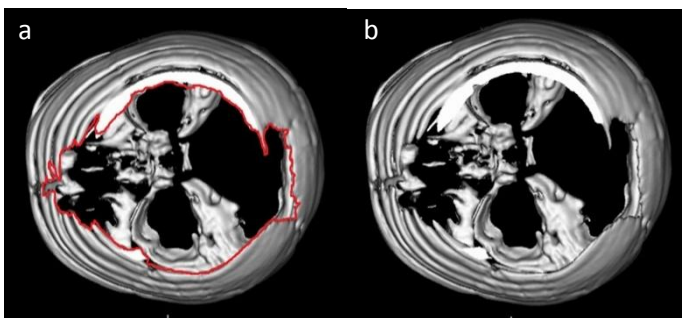


Figure 2: Three-dimensional CT scans. a & b- vertex view. b- The defect has been marked in red.

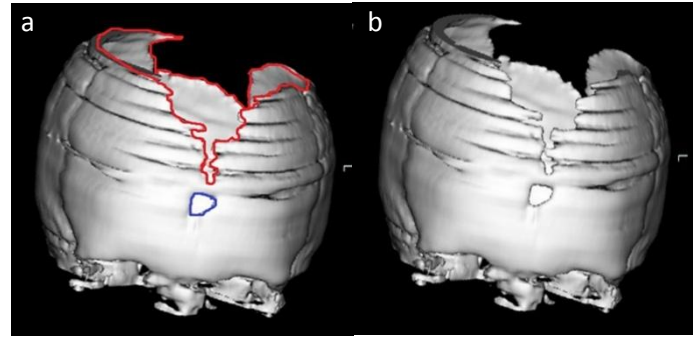


Figure 3: Three-dimensional CT scans. a- and b- anteroposterior view. b- The bigger defect has been marked in red and the second frontal defect in blue.

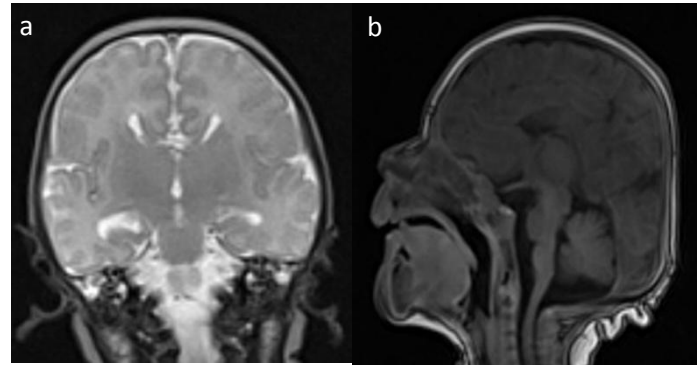


Figure 4: MRI. a- sagittal T1-weighted image showing agenesis of corpus callosum. b- coronal T2-weighted image showing crescentic shaped frontal horns and everted cingulate gyria as a result of absence of the callosal fibers.

His growths parameters are normal. Seizures have been controlled since the 4 months of age but mild developmental delay in trunk control and grasping was noted few months later; then, he was referred to a neurologic development centre, improving during rehabilitation. In the period of one year the boy also was admitted twice by non-complicated pneumonias.

The family refused further genetic workups

The boy has stable condition now and is being managed at home by teaching the parents about protection from trauma. He will continue being observed for possible cranioplasty.

DISCUSSION

The development of the skull is influenced by environmental as well as genetic factors and greatly influenced by brain growth^[2,7]. Initial neurocranial development is dependent on the formation of a membrane surrounding the neural tube. The surrounding membrane subdivides into an outer ectomeninx and an inner endomeninx. The ectomeninx produces an outer osteogenic layer, in which bone forms, and an inner dura mater. The endomeninx subdivides into the outer arachnoid and the inner pia mater. Intramembranously ossified components of the neurocranium are the bone plates of the skull such as the frontal and parietal bones. The ossification centres that develop in the membrane form the frontal, parietal, squamous temporal and squamous occipital bones. In general, each of these bones develops out of one bone centre. The parietal bone, however, develops from two bone centres that fuse with each other and subsequently function as one centre^[2]. On the other hand, neural tube defects at the cranial and spinal levels are categorized into four different types: the neural

plate remains open (anencephaly, myeloschisis); the neural tube is exteriorized (myelomeningocele and encephalocele); only the meninges is exteriorized (meningocele); and skeletal defects (cranium bifidum occultum, spina bifida occulta)^[8].

Cranium bifidum occultum is habitually the most benign type of neural tube defect, there is no herniation of cranial contents, the skull defects often close over time, there is persistent wide fontanelle and generally is asymptomatic^[5,6] but not in our case where is associated with seizures and mild neurodevelopment delay. An important element that it's necessary to keep in mind is the fact that while ossification defect persists, the brain is unprotected and very exposed to traumas.

Diagnosis is confirmed radiographically, although it may also be appreciated on palpation of the skull. In this case there is no herniation of cranial contents through the skull defect, so, cranium bifidum occultum was the proposed diagnosis.

The differential diagnosis includes paired parietal foramina which are defects in the superoposterior angles of the parietal bones via which emissary veins may pass through the calvarium; they are covered too with normal scalp and hair, and their size diminishes with age^[6]. Other differential diagnoses that should be kept in mind are: cranium bifidum with herniation of meningeal or cerebral tissue through the skull defect; acalvaria, the congenital absence of the cranial vault; anencephaly (acrania), the absence of the skull vault and of cerebral tissue; and exencephaly which is a severely malformed and exposed brain with absence of the skin and cranial vault, a forerunner of anencephaly^[4]. The documentation of facial malformations is very important too as this entity may be associated with frontonasal dysplasia^[9], a rare spectrum of anomalies that includes a variety of craniofacial defects that affect midline structures of the head and face^[10].

With relationship to our case, in the first CT scan the maximum length of the head was 93 mm and the maximum length of the defect was 89 mm (95%); the maximum width of the head: 87 mm and the width defect 72 mm (82.7%). Thirteen months later, a second CT showed a maximum length of the head of 131 mm and the maximum length of the defect was 114 mm (87%); the maximum width of the head was 106 mm and the maximum width of the defect was 77 mm (72%). There was more bone growth at the expense of the right parietal bone with minimal growth of the left parietal bone.

Treatment of such defects has been both conservative and surgical. A conservative approach has been recommended for large defects^[11], like in this case, although cranioplasty has been recommended by some neurosurgeons, if parietal bone ossification fails to fill the bone defect around the age of 13 years.

CONCLUSIONS

In this report we have described the case of a boy with severe parietal bone hypoplasia. This is the largest defect in a living patient found by the authors on the bibliographical review. Another small frontal cranium bifidum occultum, corpus callosum agenesis, lack of sutural formation of contiguous portions of left parietal and occipital bones, seizure and some degree of mental retardation were also associated. In the period of one year approximately there was minimal bone ossification. CT and MRI provided exquisite detail of both the cranial defect and the associated lesions.

Conflict of interests

The authors have none to declare

Informed consent

Was obtained from the mother.

Author contributions

Case management – M.A., V.F., M.R., C.M., C.A.; Patient follow-up – M.A., V.F., M.R.; Literature Search – V.F., C.M.; Writing – V.F.; Critical Reviews – V.F., M.A., C.M. All authors approved the final version of the manuscript.

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