Chiari IV Malformation, the Lückenschädel and Sagittal Craniosynostosis Association: Case Report and Literature Review

Associação entre malformação de Chiari IV, Lückenschädel e craniossinostose sagital: relato de caso e revisão de literatura

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Abstract

The association between Lückenschädel and craniosynostosis is unusual and unknown. Genetic origin is a possibility, representing one of many possible phenotypes for mutation. To the best of our knowledge, the association of such anomalies in a type IV Chiari malformation has never been reported before. The authors present the case of a patient with obstructive hydrocephalus, diagnosed with Chiari IV malformation associated with Lückenschädel and sagittal craniosynostosis. The Lückenschädel is the bone abnormality least commonly associated with Chiari malformation. It consists of a defect in the bones of membranous origin that form the cranial vault. This anomaly arises from periosteal dysplasia, and is characterized by rounded and irregular gaps in the skull, bound by bony ridges. Craniosynostosis is due to premature fusion of the cranial sutures and is sometimes associated with the Chiari complex of malformations. The diagnostic of Lückenschädel and craniosynostosis is done by imaging, through which the skull assumes the aspect of a hive, characteristic of Lückenschädel and it is possible to see the premature fusion of the sutures. The Lückenschädel usually does not require treatment because of spontaneous resolution; whereas, craniosynostosis
Introduction

Lückenschädel and craniosynostosis are an unusual and little-known association with possible genetic origin, representing one of many possible phenotypes for a mutation in the FGFR2 gene.\(^1\)–\(^4\) The Chiari malformations are a complex group of anatomic anomalies that usually is associated either with the Lückenschädel or to craniosynostosis. To the best of our knowledge, the association between these three anomalies has not been reported before.\(^1\),\(^2\),\(^4\)–\(^8\) In this study, the authors aim to describe this extremely rare association and to provide a review of the literature.

Case Report

The study involved a male infant, eight months old, evolving with increased head circumference and developmental delay. We performed a computed tomography (CT) scan and saw evidence of obstructive tetraventricular hydrocephalus and multiple bone defects in the cranial vault, as well as premature fusion of the sagittal suture, consistent with a diagnosis of Lückenschädel and sagittal craniosynostosis (\(\triangleright\) Figs. 1 and 2). Magnetic resonance imaging (MRI) of the skull showed a reduced size of the posterior fossa, low-setting tentorium and torcular Herophili, and herniation of the cerebellar tonsils through the foramen magnum of about 7mm (\(\triangleright\) Figs. 3 and 4). We performed a third endoscopic ventriculostomy with resolution of hydrocephalus for further programming of posterior fossa decompression and treatment of synostosis. The patient remains stable until the time of this report.

Discussion

The complex of Chiari malformations was first described in 1883 by Cleland, and graded in 1891 by the Austrian pathologist Hans Chiari. It denotes a heterogeneous group of anatomical anomalies involving the posterior cranial fossa, warrants surgery for aesthetic and functional reasons. The Chiari malformation IV only requires treatment when it results in syringomyelia or hydrocephalus.
the cerebellum, brain stem, and cranial-cervical junction, with or without changes in the lower cord. Classically, there are four accepted subtypes of malformations. Types 1 and 2 are the most prevalent, with an incidence ranging from 1 to 5 thousand births. Recently, authors have proposed subtypes 1.5 and zero, although this has not yet become widespread.

Subtype 4, presented by the authors, is uncommon, and rarely is associated with tonsillar herniation. It is characterized by a small posterior fossa, with cerebellar hypoplasia, hypoplasia of the tentorium, and low-setting torcular herophili.

The Lückenschädel is one bone abnormality that can be found in association with Arnold-Chiari (assimilation of Atlas, atlantoaxial dislocation, Klippel-Feil anomaly, platibasia, basil ar invagination), being the most unusual and generally associated with subtype 2, the most serious of the complex. This condition reaches 82% incidence in some series; however, the frequency of association with other subtypes of Chiari malformations is not known.

Also called craniolacunia, lacunar skull, or fenestrated skull, the Lückenschädel consists of a defect in the bones of membranous origin that form the cranial vault. This anomaly is due to periosteal dysplasia, and is characterized by rounded and irregular gaps in the skull, bounded by bony ridges. Pathologically such failures are completely devoid of bony structure formed by only a membranous diaphragm of periosteum and dura mater. The same defect is not found in the skull base bones, which have cartilaginous origin.

The association between Lückenschädel and craniosynostosis is already known, but uncommon, occurring in 10% of cases of craniosynostosis. It is likely caused by mutations in the FGFR2 gene (fibroblast growth factor receptor 2) involved in the genesis of craniosynostosis, which have been found in cases of craniolacunia, raising suspicion that mutations in different exons would be responsible for different phenotypes. This would explain the association in the present case.

The Lückenschädel and craniosynostosis diagnoses are done through imaging studies, both X-ray and computed tomography, in which is possible to see the bone defects and fusion of the cranial sutures. When examining X-rays or CT with 3D

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**Fig. 2** 3-D reconstruction CT scan depicts the multiple bone defects of Lückenschädel.

**Fig. 3** Sagittal T1W MRI depicts the reduced size of the posterior fossa, low-setting torcular herophili, and herniation of the cerebellar tonsils through the foramen magnum.

**Fig. 4** Sagittal T1W MRI depicts the reduced size of the posterior fossa and low-setting tentorium.
reconstruction, the skull assumes the aspect of a hive, which is a
typical characteristic of Lückenschädel.1–4,11–13,15,16,21,23,24

This anomaly usually does not require treatment because the
ossification tends to normalize between the fourth and
sixth month of life, and does not directly reflect in brain
development.1,3,4 Given its spontaneous and early resolution,
it is not seen in adults, which contributes to the low rate
diagnosis of this condition.1,3,4 The Chiari malformation
type IV also tends to be asymptomatic, except for the few
cases where there may be tonsillar herniation, determining
syringomyelia and/or hydrocephalus. In this case, neurosur-
gical intervention is required, as in the present case.5–7

Craniosynostosis implies surgery for aesthetic and functional
purposes and should be performed as early as possible
after diagnosis.23,24

The association between Chiari 4 malformation, Lück-
enshchädel and craniosynostosis is extremely unusual, being
so little known and diagnosed; however, newborn imaging
studies may increase the diagnostic rate of these diseases
and better guide the medical care of such cases.

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