

Chiari malformation in nonsyndromal single craniosynostosis—Much Ado about Nothing?

M. Engel · G. Castrillón-Oberndorfer · J. Hoffmann ·
B. Orakcioglu · S. Rohde · R. Seeberger · C. Freudlsperger

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Abstract

Background The significance of Chiari malformation in nonsyndromal-isolated craniosynostosis is still not well documented. Hence, in the present study we investigated the incidence of Chiari malformation in a larger series of patients with nonsyndromic-isolated single-suture craniosynostosis over a 9-year period using preoperative magnetic resonance imaging (MRI).

Methods Of 215 children who had undergone surgery for nonsyndromic-isolated craniosynostosis, 89 cases (41.4 %) had MRI prior to surgery. All MRIs were screened for Chiari malformation.

Results Only one patient (1.1 %) with isolated lambdoid synostosis showed Chiari malformation preoperatively, which was defined as a cerebellar tonsillar descent greater than 5 mm below the foramen magnum. However, no clinical symptoms were associated with Chiari malformation in this patient.

Conclusions As Chiari malformation is more likely to be associated with syndromic craniosynostosis, nonsyndromic bilateral coronal synostosis, or synostosis of the lambdoid suture, a general use of MRI as a screening tool for Chiari malformation should not be recommended for patients with

nonsyndromic-isolated craniosynostosis who lack clinical symptoms.

Keywords Chiari malformation · Craniosynostosis · Scaphocephaly · Craniofacial

Introduction

Chiari malformation (CM) is characterized by downward herniation of neural tissue through the foramen magnum with a displacement of the cerebellar tonsils [7]. The association between CM and craniosynostosis has been first noted by Saldino et al. in a description of a child with Pfeiffer syndrome [9]. Since that time, and until the advent of MRI evaluation in pediatric neurosurgical practice, several cases have been reported in the literature [2, 4, 5, 11]. The pathogenesis of this malformation is still discussed controversially and optimal treatment options are not well established [2, 11, 12]. CM occurs in patients with both syndromic and nonsyndromic forms of craniosynostosis, however, patients with syndromal craniosynostosis including Crouzon, Apert, or Pfeiffer syndrome are frequently associated with CM in over 50 % of all cases [1, 3, 11] (Fig. 1). In addition, nonsyndromic oxycephaly is found in two-thirds of all cases [2]. Besides the association of syndromal craniosynostosis or nonsyndromic bilateral coronal synostosis and CM, there are several reports of CM in nonsyndromic-isolated craniosynostosis [7, 8, 11, 13]. However, clinical studies of CM in nonsyndromal-isolated craniosynostosis are rare, and the incidence of this association is still not well documented [7].

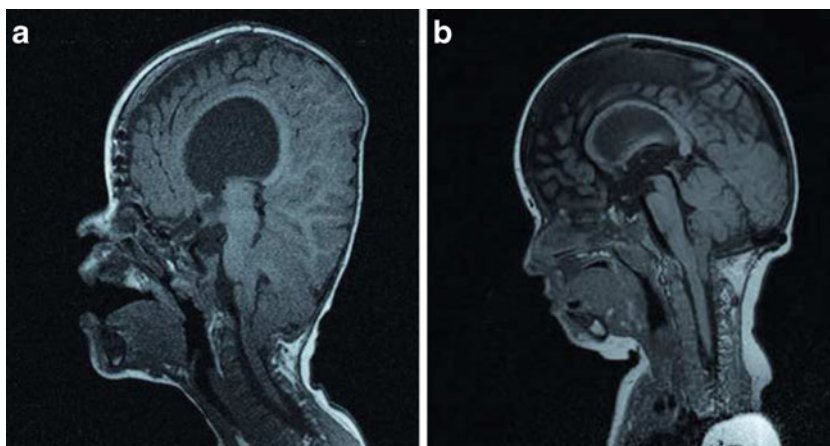
In the present study, we investigated the incidence of CM in patients with nonsyndromic-isolated single-suture craniosynostosis over a 9-year period using preoperative magnetic resonance imaging (MRI).

M. Engel (✉) · G. Castrillón-Oberndorfer · J. Hoffmann ·
R. Seeberger · C. Freudlsperger
Department of Oral and Maxillofacial Surgery,
University Hospital Heidelberg,
Im Neuenheimer Feld 400,
69120 Heidelberg, Germany
e-mail: michael.engel@med.uni-heidelberg.de

B. Orakcioglu
Department of Neurosurgery, University Hospital Heidelberg,
Heidelberg, Germany

S. Rohde
Department of Neuroradiology, University Hospital Heidelberg,
Heidelberg, Germany

Fig. 1 Examples for Chiari malformation in syndromal craniosynostosis. Sagittal MR image of an asymptomatic patient with Apert syndrome showing cerebellar tonsillar descent with crowding at foramen magnum (a). Sagittal MR image in a symptomatic patient with Crouzon syndrome showing cerebellar tonsillar descent to 14 mm below the foramen magnum (b). A successful CM decompression was performed in the follow-up



Patients and methods

For this retrospective, non-blinded, case-review study, the study population consisted of all patients with nonsyndromic-isolated craniosynostosis who had MRI diagnostic prior to surgery between January 2001 and December 2009 in the Department of Oral and Maxillofacial Surgery. This study has been approved by the institution's ethics committee in accordance with the ethical standards of the 1964 Declaration of Helsinki. All patients or their legal guardian gave their written informed consent prior to their inclusion in the study. From January 2001 to December 2005, MRI studies were performed as a standard diagnostic tool according to our former diagnostic algorithm for nonsyndromic-isolated craniosynostosis ($n=69$). Since then, we only performed MRI when any clinical signs of increased ICP (e.g., papilledema) were present ($n=9$). The remaining MRIs have been already performed by the referring physician or clinic at time of referral ($n=11$).

MRI studies were performed with a 1.5-Tesla MR unit. We obtained T1-weighted spin echo images in sagittal and axial planes and T2-weighted spin echo images in axial and coronal planes. Patients were sedated during the MR investigation. All MRI images were reviewed by an experienced colleague of the Department of Neuroradiology. Sagittal MR images were evaluated for the presence of CM: the descent of the tonsils was measured by calipers from the foramen magnum. In line with others, the arbitrary cut-off point for definition of CM was a cerebellar tonsillar descent greater than or equal to 5 mm below the foramen magnum [11]. For each case, we recorded demographic and clinical information and characteristics of the craniosynostosis.

Results

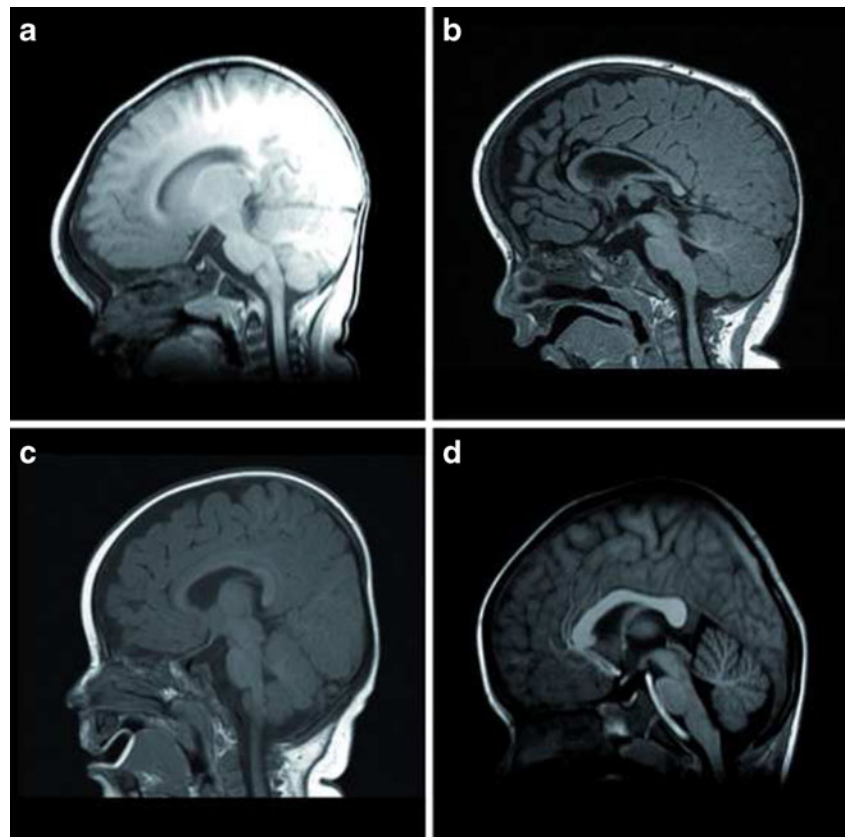
We identified 215 children who had undergone surgery for nonsyndromic-isolated craniosynostosis in the specified period. Altogether, 89 children (41.4 %) had MRI prior to surgery. Of these patients, 42 children (47.2 %) were

diagnosed for isolated sagittal synostosis, 14 (15.7 %) for isolated unilateral coronal synostosis, 29 (32.6 %) for isolated metopic synostosis, and 4 (4.5 %) for isolated lambdoid synostosis. Furthermore, known predisposing factors for CM like hypophosphatemic rickets could not be identified in our series [7]. The average age of our patients at time of MRI was 11.6 months (standard deviation 4.2 months). In the majority of all cases, surgery was performed 1 week after MRI. According to the surgical reports, the diagnosis of craniosynostosis was confirmed intraoperatively in all cases. In the present study, 88 patients (98.9 %) showed no signs of CM in the preoperative MRI scan (Fig. 2). On the other hand, the incidence of CM in association with nonsyndromal single craniosynostosis was only 1.1 %. Of the 89 patients, only one patient with isolated lambdoid synostosis showed a cerebellar tonsillar descent greater than 5 mm below the foramen magnum (Fig. 3). This patient showed no clinical symptoms, which could have been attributed to CM.

Discussion

The association between CM and syndromic and nonsyndromic multisutural craniosynostosis has been recognized for many years [9]. Previous studies demonstrated that the incidence of CM was as high as 70 % in Crouzon's syndrome, 75 % in nonsyndromic oxycephaly, 50 % in Pfeiffer's syndrome, and 100 % in Kleeblattschädel deformity and other rare cranio-facial syndromes [2]. Notably, the majority of these cases showed the presence of a multisutural craniosynostosis, syndromic or nonsyndromic, with a predominant involvement of the posterior aspect of the skull [2]. However, cases of nonsyndromic-isolated craniosynostosis without predictive factors were rare. Hence, we investigated the incidence of CM in patients with nonsyndromic-isolated single-suture craniosynostosis. In our series, metopic synostosis was not found to be associated with CM. The same was also reported by Leikola et al. [7] who

Fig. 2 Sagittal MR images of four patients with nonsyndromic-isolated craniosynostosis. **a** Isolated metopic synostosis. **b** Isolated sagittal synostosis. **c** Isolated coronal synostosis. **d** Isolated lambdoid synostosis. All patients showed no signs of Chiari malformation, which was defined as a displacement of the cerebellar tonsils more than 5 mm through the foramen magnum



showed in a series of 124 patients with nonsyndromic-isolated craniosynostosis, no association between isolated metopic synostosis and CM. However, the authors did not provide the number of patients with isolated metopic synostosis.

Similar results were reported by Strahle and colleagues. In their large series of 71 patients with nonsyndromic-isolated metopic synostosis, no patient was associated with CM as investigated using preoperative MRI [11]. Surprisingly, these findings were in contrast to the results reported by Tubbs and colleagues. In their prospective evaluation of 50 children with

simple metopic ridges without other signs of trigonocephaly, a 30 % incidence of CM was described [13]. The authors postulated that this might be related to the reduced volume of the anterior fossa, however, the complete reasons remain unclear. As the authors did not provide any information about the use of MRI for the diagnosis of CM, this unexpected high incidence of CM might be considered critically. CM was also found in rare cases of isolated sagittal and unilateral coronal synostosis. Strahle et al. investigated CM in 183 isolated sagittal and 80 isolated unilateral coronal craniosynostosis (syndromic and nonsyndromic). In that collective, only three

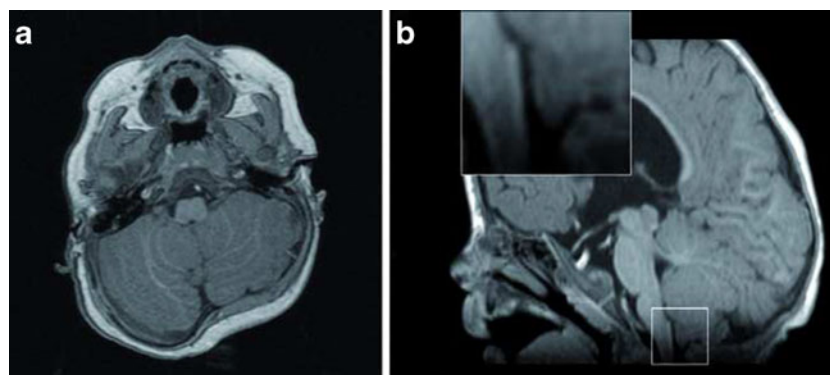


Fig. 3 Patient with isolated lambdoid synostosis and manifestation of Chiari malformation. Axial preoperative MRI scans showing typical skull deformity caused by isolated lambdoid synostosis on the left side (**a**). Sagittal preoperative MRI scans indicating Chiari malformation

defined as a downward herniation of neural tissue through the foramen magnum with a displacement of the cerebellar tonsils more than 5 mm. Insert at 3x magnification

patients with single-suture, nonsyndromic sagittal craniosynostosis and no patients with nonsyndromic unilateral coronal synostosis showed associated CM [11]. Leikola et al. report a 5.6 % overall incidence of CM in nonsyndromic single-suture synostosis [7]. In their series, two patients with coronal and five with sagittal synostosis were identified. However two of the five patients with isolated sagittal synostosis showed a hereditary hypophosphatemic type of rickets as a known predisposing factor for CM [7]. The reason for the association of CM and hypophosphatemic rickets is considered to be due to bony overgrowth of the posterior cranial fossa with succeeding reduced volume of the posterior fossa [7]. Cinalli et al. reported about the important role of the lambdoid suture in CM [2]. Lambdoid suture involvement is predictive for CM formation even in patients with multisuture or syndromic forms of craniosynostosis. The progressive fusion of the lambdoid suture (associated or not with closure of cranial base synchondroses) produces alteration in the skull base and stenosis of the jugular foramina. The first result would be a small posterior fossa with consequent herniation of the cerebellum into the cervical canal during the phase of rapid neural growth in the very first months of life. So, the frequent association of lambdoid synostosis with CM is well established [11]. Strahle et al. found CM in five of nine cases with isolated lambdoid synostosis. In contrast to that, we found only one patient with lambdoid synostosis and association with CM. A reason therefore could be the small number of patients with isolated lambda synostosis ($n=4$ patients) in our study due to the very low incidence of 1 to 100,000 cases per live births. A similar result was reported by Leikola et al., who could not identify a patient with isolated lambdoid synostosis and CM [7].

On the basis of the previously reported studies and our own experience, we believe that CM is frequently associated with syndromic and multisuture synostosis, however, has a low incidence in nonsyndromic-isolated craniosynostosis. If present in nonsyndromic-isolated craniosynostosis, CM usually shows an asymptomatic clinical course and often presents as an accidental finding in MRI. Generally, most surgeons agree that CM should not be treated unless it is symptomatic or, in some cases, associated with a spinal syrinx. However, the indications for surgical treatment of CM may be different in patients with associated craniosynostosis as CM decompression might be performed at the time of a planned craniosynostosis repair. Interestingly, several groups recommend posterior fossa expansion surgery as the treatment of choice for all cases of CM identified prior to craniosynostosis correction, even in the absence of symptoms [2, 10, 14].

Eighty-nine of 215 children (41.4 %) who had undergone surgery for nonsyndromic-isolated craniosynostosis in the specified period had MRI prior to surgery, compared to the study of Strahle and colleagues, where only 14.3 %

of patients with nonsyndromic single-suture synostosis received preoperative MRI diagnostic [11]. This relatively high percentage of MRI in our patients was due to our former diagnostic algorithm for nonsyndromic-isolated craniosynostosis (January 2001 to December 2005), where MRI was routinely performed. On the other hand, 126 patients (58.6 %) with nonsyndromic-isolated craniosynostosis had no MRI prior to surgery. However, our documentation revealed no symptoms related to clinical signs of CM. We suppose if there had been clinical symptoms suspicious for CM, these patients had been referred and imaged. We also assume that some rare asymptomatic cases could be found, if these 126 patients were screened for CM. In addition, besides the detection of CM in craniosynostosis, MRI is an excellent technique for the diagnosis of associated diseases of the cerebrum like, e.g., midline anomalies, lesions of the parenchyma, intracranial herniation, and hydrocephalus [6].

For our clinical routine, we are in line with Strahle et al., who recommends screening of patients with syndromic craniosynostosis or with lambdoid synostosis prior to surgical correction using MRI. From our point of view, a routine screen of asymptomatic patients with isolated single-suture craniosynostosis at locations other than the lambdoid suture is not necessary, considering the possible risks of sedation required for MRI in children.

Conclusions

As CM, symptomatic or asymptomatic, is a regular finding in patients with syndromal craniosynostosis, with nonsyndromic bilateral coronal synostosis or with isolated lambdoid craniosynostosis, those patients should generally be scanned for CM using MRI.

However, association between CM and nonsyndromic-isolated craniosynostosis at locations other than the lambdoid suture seems to be too rare to recommend a general use of MRI as screening tool for CM, especially as the majority of patients with CM in nonsyndromic-isolated craniosynostosis lack clinical symptoms.

Conflicts of interest None.

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