Growing Skull Defect in an Infant with a Rare Combination of a Foramen Parietale Permagna and an Atretic Cephalocele

Miriam Ratliff *, Andreas Unterberg and Heidi Bächli

Department of Neurosurgery, Heidelberg University, Germany

Abstract: Here we present a case of a 2-month-old child with an atretic encephalocele and large persistent parietal foramina. The course was unusual in that the parietal foramina significantly increased in size over a relatively short time.

At the age of three months the child required surgery because of the increasing skull defect. During surgery the cause of the growing skull defect was revealed as a medial atretic encephalocele with enlarged parietal foramina.

Large parietal foramina are a rare clinical entity with a prevalence ranging from 1:15.000 to 1:25.000. The skull defect is usually identified on physical examination and confirmed radio graphically.

We assume that the mechanism underlying the growing bone defect is identical to that of a growing skull fracture. To our knowledge this is the only reported case of an infant with a growing skull defect requiring surgery due to an atretic encephalocele protruding through a growing parietal foramina.

Keywords: leptomeningeal cyst, foramen parietale permagna, atretic encephalocele.

1. INTRODUCTION

The existence of small parietal foramina (1-2mm) is considered a common normal variant; they are skull perforations for passage of emissary veins. These veins connect occipital veins with the superior sagittal sinus and an anastomosis between the middle meningeal arteries and the occipital arteries. They are typically located near the parietal eminence and occur uni- or bilaterally in 65% of the population [4, 13].

Enlarged parietal foramina are also known as fenestrae parietalis symmetrical, foramina parietalia permagna, giant parietal foramina and Catlin marks. In contrast to normal small parietal foramina enlarged parietal foramina are a hereditary condition. Goldsmith described 5 generations of the Catlin family with enlarged parietal foramina [7]. Transmission is autosomal dominant with incomplete penetrance; responsible genes have been identified [4, 10, 21].

Despite being located similar to the small parietal foramina, they are believed to be a developmental anomaly of parietal bone ossification. During development they first exist as a single ossification defect involving both parietal bones [4, 21, 24]. The ossification gap is eventually divided into two foramina by parasagittal islands of ossification forming a midline bridge during the fifth month of gestation [6, 21]. A persisting central lack of ossification may present as enlarged posterior fontanel termed cranium bifidum [30].

Besides encephaloceles, myelomeningoceles, craniofacial and skeletal anomalies have infrequently been associated with enlarged parietal foramina [29].

2. CASE REPORT

Patient history and clinical exam. The female patient was first presented to our department at 2 months of age. She was born mature at gestational age 38 weeks, with a birth weight of 2710 g. The height at birth (46cm) and head circumference (33.5cm) was within the percentiles. The pre-natal history was unremarkable. Already at birth a soft subscalp midline swelling was noted parietooccipital, over time the lesion was documented to grow in size. When first introduced to our department the patient showed wide sutures. The fontanel was not elevated. The described lesion on the scalp was about 5x2cm in diameter. The parietooccipital mass was soft and fluctuating, with hairy skin overlying it (Figure **1**). The patient had no other neurological symptoms, physical examination was normal, and psychomotor development was parallel within the age of the child. During a follow up visit a month later the lesion had grown to 7x3cm.

Neuroimaging studies, operative treatment, and outcome. An MRI showed a complex syndrome with a small parietal paramedian encephalocele surrounded by a subdural collection of cerebrospinal fluid. As typically seen in parietooccipital encephalocele, a “peaking” tentorium and a vertical positioning of the superior sagittal sinus was revealed (Figure **1**) [23]. An

*Address correspondence to this author at the Miriam Ratliff, Department of Neurosurgery, University Medical Center Mannheim, University of Heidelberg, Theodor-Kutzer-Ufer 1-3, 68167 Mannheim, Germany; Tel: +49 0621 383-2750; Fax: +49 621 383-2004; E-mail: miriam.ratliff@umm.de

E-ISSN: 2311-8687/16

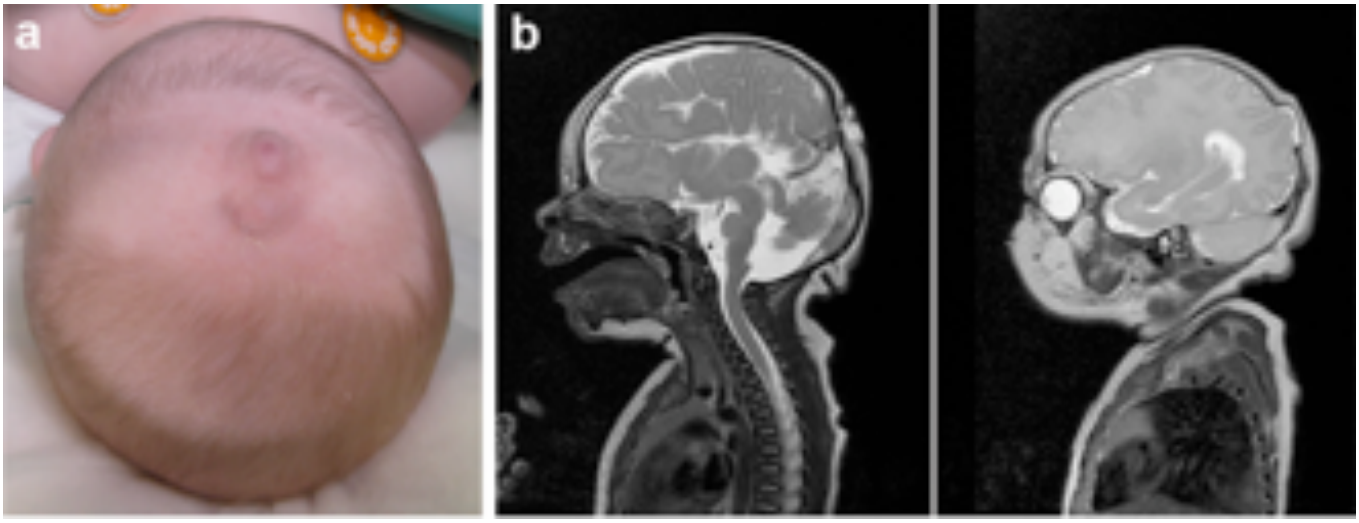

Figure 1a: Subscalp parietooccipital swelling without a skin lesion. **b:** MRI imaging revealed a complex syndrome and a paramedian encephalocele. Sagittal T2 MRI showing a "peaking" tentorium and a vertical positioning of the superior sagittal sinus. A high falx/tentorial junction was observed.

abnormally positioned straight sinus was suspected on sagittal MR images, identifiable as a linear flow void extending posteriorly and superiorly within the posterior interhemispheric fissure to the base of the subscalp lesion. A high falx/tentorial junction associated with prominence of the subjacent superior cerebellar cistern was seen. The MRI also revealed an agenesis of the corpus callosum and a hypoplastic vermis.

Because of the rapid increase in size of the subscalp lesion surgical treatment was indicated. During surgery the lesion was identified as atretic encephalocele protruding through enlarged parietal foramina measuring 3x2 and 2x2.5cm. The brain tissue was elevated above bone level and lacked a dura cover. Surgical treatment involved dura repair using an artificial dura substitute (DuraGen Plus™ Integra LifeSciences Corporation, Plainsboro, USA) and cranioplasty using surrounding normal skull. Treatment was chosen as suggested for growing skull fractures (GSF) combining closure of the dural rent with autologous cranioplasty [2, 9, 27] (Figure **2**). Follow-ups over the following years revealed a good aesthetic outcome, with no indication of scull or dural defect.

3. DISCUSSION

The incidence of atretic encephaloceles is described to be 4-17% [18]. The most frequent location is parietal (40-50%) [32]. Supposedly the underlying pathological mechanism is a faulty neural tube closure during embryogenesis.

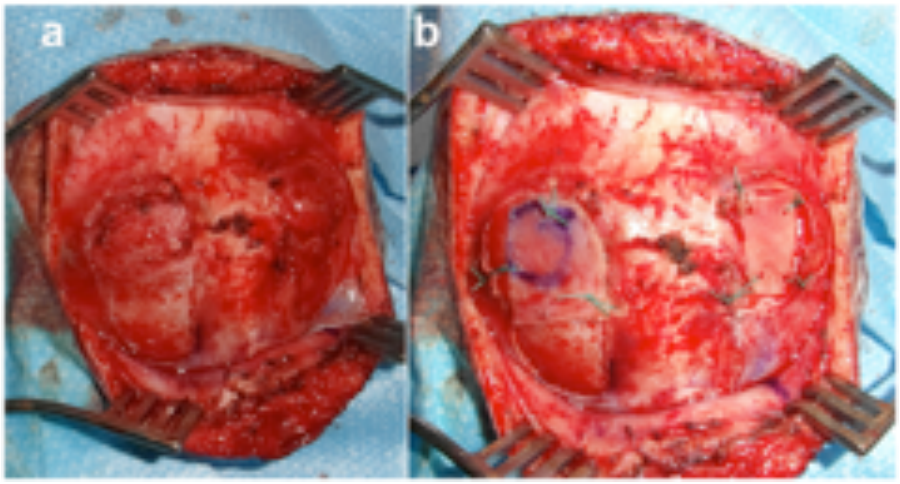

Figure 2: Operative site. During surgery the enlarged parietal foramina were revealed (left image). Surgical treatment involved duraplasty and autologous cranioplasty (right image).

It has previously been suggested that aberrant vascular evolution during fetal development may affect cerebrovascular, brain or skull development [26]. Abnormalities of venous drainage are recognized in association with atretic encephaloceles / meningoceles and are also present in a very similar pattern in enlarged parietal foramina [3, 18, 23]. In fact Reddy *et al*. described a case of enlarged parietal foramina that also had a previously undiagnosed atretic occipital encephalocele [26]. Aoyagi *et al*. presented a case of symmetrical parietal meningoceles associated with enlarged parietal foramina [1]. In both cases abnormal venous anatomy was present. Similar venous anomalies in atretic parietal encephaloceles and enlarged parietal foramina lead to the hypothesis that the underlying developmental mechanism is related and that possibly enlarged parietal foramina represent the benign end of the same developmental spectrum [5]. Typically atretic parietal encephaloceles are associated with skull defects in the midline and frequently present with a subcutaneous midline cyst or nodule. The overlying scalp shows abnormalities whereas in parietal foramina the overlying scalp is always normal [5]. There was no scalp lesion in our case. James and Lassman were the first to report atretic encephaloceles in 1972 [5].

The first report of a GSF, an enlarging cranial defect arising from a linear skull fracture was given by How ship in 1816 [14]. Several reports followed, applying various terms such as leptomeningeal cyst or the aforementioned growing skull fracture. The incidence of growing skull fractures was reported by Arseni to be less than 0.05% in cases with a traumatic skull fracture [2]. In the series of Muhonen *et al*. an incidence of 0.2% was documented [19]. If only pediatric cases of skull fractures would be considered we assume that the incidence would be significantly higher as the majority of cases with GSF have been seen in patients less than 1 year of age. GSFs in patients older than 3 years of age are quite rare [17, 28]. More frequently children under 3 years of age are affected because the dura is more closely adherent to the bone rendering it more susceptible to tearing during fracture. Furthermore we hypothesize that intermittent increased intracerebral pressure (ICP) during periods of crying contributes to the progression of GSFs in neonates and young children. Protruding meanings trap subarachnoid fluid in the sac. The shape of the bony vault is primarily determined by the expanding brain [20, 32]. In cases of GSF the vault becomes distorted. Transmission of brain pulsations gradually cause skull erosions, remodeling of the surrounding bone and enlargement of the sac [11, 12]. The pressure of the unrestrained brain initiates progressive bone resorption allowing enlarging cerebral hernia.

The fast clinical progression of the skull defect as seen in our case is not that unusual. Some authors report development of a growing skull fracture in as little as three months after trauma [17]. It has been described that the usual time period from injury to presentation with GSF is typically less than 3 years [28].

Experimental studies have examined the underlying pathological mechanism of GSFs. Keener found in 1958 that a dural laceration was necessary to produce a GSF. He also found that the development of an enlarging fracture was 2.5 times higher if the pia and the arachnoid was injured [16]. Ramamurthi and Kalyanaraman demonstrated in a study with 6 to 17 day old puppies that a dura laceration underneath a craniotomy involving the arachnoid is sufficient for development of a growing fracture [25]. Goldstein, *et al*. confirmed these findings in an investigation on 4 to 8 week old puppies. According to his data the arachnoid as well as the dura must be interrupted to support a growing skull fracture. Additional brain injury had no effect on the incidence of growing skull fractures [8]. Whereas most cases of GSF occur as a consequence of severe head trauma involving brain injury the pathomechanism of the case presented here is likely to be very similar to the experimental settings without parenchymal affection. It is not known what minimal size of a dural lesion is required to initiate the formation of a growing skull defect. But reports on “GSF” after craniofacial surgeries in children under the age for three years suggest that dural defects can be very small and still lead to growing skull defects during the postoperative cause. Frequently a dural laceration went unnoticed during open craniofacial surgeries assuming that the dural tear must have been minor.

This case parallels reported cases of children with GSF in such that GSF in general have a strong tendency to occur and to progress rapidly in size in infancy. Pathological findings involve abnormality of dura, progressive bone defects, and increasing brain herniation. An identical surgical procedure that has been successfully applied in the past in children with GSF, involving duraplasty and autologous cranioplasty was effective in treatment.

We believe for the reasons discussed above, that our patient has fundamentally the same disorder as children with growing skull fractures after severe head

trauma. In summary this case concurs with Taveras and Ransohoff that a dural tear in addition to an outward driving force such as a growing brain, hydrocephalus, cerebral edema or neoplasm predispose to a growing skull defect [28].

SUMMARY

In summary, to the best of our knowledge this is the first described case of a patient with enlarged parietal foramina associated with an atretic encephalocele that required surgery as a neonate due to a growing scull defect. If GSFs are considered, the diagnosis of an enlarging bone and dura defect is not difficult to confirm in a case of a fluctuating, progressive, soft subscalp mass. A leptomeningeal cyst can be diagnosed with appropriate neuroimaging. If left untreated, brain parenchyma eventually may herniate through the dural laceration resulting in potential neurological symptoms such as hemiparesis or the development of seizures.

As awareness of venous anomalies is imperative for surgical management, we recommend MRI as a diagnostic method of choice. We agree with Winston *et al*., who recommended that dural defects in children should be repaired as soon as possible, because of the progressive character of these lesions, and because of the risk of secondary injury to brain [31]. Early surgery prevents seizures and motor deficits that are prone to occur during the enlargement of these dural defects if they remain untreated.

CONFLICT INTEREST

The authors report no conflict of interest concerning the materials or methods used in this study or the findings specified in this paper.

REFERENCE

[1] Aoyagi M, Matsushima Y, Takei H, Okada K and Inaba Y. Parietal foramina complicated by meningocele. Childs Nerv Syst 1985; 1: 234-237. https://doi.org/10.1007/BF00270769

[2] Arseni C and Ciurea AV. Clinicotherapeutic aspects in the growing skull fracture. A review of the literature. Childs Brain 1981; 8: 161-172.

[3] Bartels RH, Merx JL and van Overbeeke JJ. Falcine sinus and occipital encephalocele: a magnetic resonance venography study. J Neurosurg 1998; 89: 738-741. https://doi.org/10.3171/jns.1998.89.5.0738

[4] Fein JM and Brinker RA. Evolution and significance of giant parietal foramina. Report of five cases in one family. J Neurosurg 1972; 37: 487-492. https://doi.org/10.3171/jns.1972.37.4.0487

[5] Fink AM and Maixner W. Enlarged parietal foramina: MR imaging features in the fetus and neonate. Am J Neuroradiol 2006; 27: 1379-1381.

[6] Golabi M, Carey J and Hall BD. Parietal foramina clavicular hypoplasia. An autosomal dominant syndrome. Am J Dis Child 1984; 138: 596-599. https://doi.org/10.1001/archpedi.1984.02140440080022

[7] Goldsmith PG. The Pathology and Treatment of Chronic Catarrhal Deafness. Can Med Assoc J 1922; 12: 293-299. https://doi.org/10.1288/00005537-192209000-00010

[8] Goldstein F, Sakoda T, Kepes JJ, Kavidson K and Brackett CE. Enlarging skull fractures: an experimental study. J Neurosurg 1967; 27: 541-550. https://doi.org/10.3171/jns.1967.27.6.0541

[9] Goldstein FP, Rosenthal SA, Garancis JC, Larson SJ and Brackett CE. Jr Varieties of growing skull fractures in childhood. J Neurosurg 1970; 33: 25-28. https://doi.org/10.3171/jns.1970.33.1.0025

[10] Halbertsma T. Fenestrae Parietales Symmetricae. Arch Dis Child 1940; 15: 115-120. https://doi.org/10.1136/adc.15.82.115

[11] Halliday AL, Chapman PH and Heros RC. Leptomeningeal cyst resulting from adulthood trauma: case report. Neurosurgery 1990; 26: 150-153. https://doi.org/10.1227/00006123-199001000-00025

[12] Haworth CS and Pobereskin LH. Middle fossa arachnoid cyst eroding into the middle ear: case report. Neurosurgery 1990; 26: 154-155. https://doi.org/10.1227/00006123-199001000-00026

[13] Hollender L. Enlarged parietal foramina. Oral Surg Oral Med Oral Pathol 1967; 23: 447-453. https://doi.org/10.1016/0030-4220(67)90536-1

[14] Howship J. Practical observations in surgery and morbid anatomy. London Engans Longman 1816; pp 494.

[15] James CC and Lassman LP. Spina bifida occulta. James CC, Lassman LP: Spinal Dysraphism, 1st edn. Butterworths, London 1972; 89-97.

[16] Keener EB. An experimental study of reactions of the dura mater to wounding and loss of substance. J Neurosurg 1959; 16: 424-447. https://doi.org/10.3171/jns.1959.16.4.0424

[17] Kingsley D, Till K and Hoare R. Growing fractures of the skull. J Neurol Neurosurg Psychiatry 1978; 41: 312-318. https://doi.org/10.1136/jnnp.41.4.312

[18] Martinez-Lage JF, Sola J, Casas C, Poza M, Almagro MJ and Girona DG. Atretic cephalocele: the tip of the iceberg. J Neurosurg 1992; 77: 230-235. https://doi.org/10.3171/jns.1992.77.2.0230

[19] Muhonen MG, Piper JG and Menezes AH. Pathogenesis and treatment of growing skull fractures. Surg Neurol 1995; 43: 367-372. https://doi.org/10.1016/0090-3019(95)80066-P

[20] Mücke R and Meyer-Glauner M. Duraspannung und Schädelwachstum. Acta Neurochir 1977; 37: 281-291. https://doi.org/10.1007/BF01402132

[21] Otsubo Y, Sato H, Sato N and Ito H. Cephaloceles and abnormal venous drainage. Childs Nerv Syst 1999; 15: 329-332. https://doi.org/10.1007/s003810050405

[22] Pang D and Lin A. Symptomatic large parietal foramina. Neurosurgery 1982; 11: 33-37. https://doi.org/10.1227/00006123-198207010-00007

[23] Patterson RJ, Egelhoff JC, Crone KR and Ball WS Jr. Atretic parietal cephaloceles revisited: an enlarging clinical and imaging spectrum? Am J Neuroradiol 1998; 19: 791-795.

[24] Preis S, Engelbrecht V and Lenard HG. Aplasia cutis congenita and enlarged parietal foramina (Catlin marks) in a family. Acta Paediatr 1995; 84: 701-702. https://doi.org/10.1111/j.1651-2227.1995.tb13735.x

[25] Ramamurthi B and Kalyanaraman S. Rationale for surgery in growing fractures of the skull. J Neurosurg 1970; 32: 427-430. https://doi.org/10.3171/jns.1970.32.4.0427

[26] Reddy AT, Hedlund GL and Percy AK. Enlarged parietal foramina: association with cerebral venous and cortical anomalies. Neurology 2000; 54: 1175-1178. https://doi.org/10.1212/WNL.54.5.1175

[27] Scarfo GB, Mariottini A, Tomaccini D and Palma L. Growing skull fractures: progressive evolution of brain damage and effectiveness of surgical treatment. Childs Nerv Syst 1989; 5: 163-167. https://doi.org/10.1007/BF00272120

[28] Taveras JM and Ransohoff J. Leptomeningeal cysts of the brain following trauma with erosion of the skull; a study of seven cases treated by surgery. J Neurosurg 1989; 10: 233-241. https://doi.org/10.3171/jns.1953.10.3.0233

[29] Warkany J. Syndromes. Am J Dis Child 1971; 121: 365-370. https://doi.org/10.1001/archpedi.1971.02100160035001

[30] Wilkie AOM and Mavrogiannis LA. Enlarged Parietal Foramina. Pagon RA, Adam MP, Bird TD, Dolan CR, Fong CT, Smith RJH, et al. (eds): GeneReviews(R). Seattle (WA) 1993.

[31] Winston K, Beatty RM and Fischer EG. Consequences of dural defects acquired in infancy. J Neurosurg 1983; 59: 839-846. https://doi.org/10.3171/jns.1983.59.5.0839

[32] Yokota A, Kajiwara H, Kohchi M, Fuwa I and Wada H. Parietal cephalocele: clinical importance of its atretic form and associated malformations. J Neurosurg 1988; 69: 545-551. https://doi.org/10.3171/jns.1988.69.4.0545

[33] Young RW. The influence of cranial contents on postnatal growth of the skull in the rat. Am J Anat 1959; 105: 383-415. https://doi.org/10.1002/aja.1001050304

Received on 08-12-2016 Accepted on 19-12-2016 Published on 31-12-2016

DOI: http://dx.doi.org/10.12974/2311-8687.2016.04.02.2