PORTUGUESE JOURNAL OF PEDIATRICS

PERMANYER

Check for updates

CASE REPORT

Enlarged parietal foramina: a case report

Mariana Gaspar[1a], Susana Moleiro[1b], Carla Conceição[2c], and Amets Sagarribay[3d]*

[1]Serviço de Pediatria, Departamento da Mulher e da Criança, Hospital de Santarém, Santarém; [2]Serviço de Neurorradiologia, Hospital Dona Estefânia, Centro Hospitalar Universitário de Lisboa Central, Lisbon; [3]Unidade de Neurocirurgia Pediátrica, Hospital Dona Estefânia, Centro Hospitalar Universitário de Lisboa Central, Lisbon. Portugal

ORCID: [a]0000-0002-5239-5820; [b]0009-0009-4526-5049; [c]0000-0003-1430-1122; [d]0000-0003-2280-8417

Abstract

Enlarged parietal foramina is a rare entity that results from late or incomplete ossification, ending as large foramina in the parietal bone. Usually, affected children are asymptomatic but may present with severe headache, vomiting or predisposition for epilepsy. This case aims to raise awareness of a rare clinical entity that can be manifested by a persistently opened posterior fontanelle. The prognosis is mostly benign.

Keywords: Parietal foramina. Posterior fontanelle. Autosomal dominant. Benign.

Foramina parietalia permagna: caso clínico

Resumo

Foramina parietais alargados representam uma entidade rara que resulta de ossificação tardia ou incompleta, culminando num defeito ósseo de grandes dimensões no osso parietal. Geralmente, as crianças afetadas são assintomáticas, podendo no entanto apresentar-se com cefaleia, vómitos ou predisposição para epilepsia. Este caso tem como objetivo alertar para uma entidade clínica rara que pode manifestar-se por uma fontanela posterior persistentemente aberta. O prognóstico é geralmente benigno.

Palavras-chave: Foramina parietais. Fontanela posterior. Autossómica dominante. Benigno.

The authors report the case of a 2-day-old girl discharged from a newborn nursery to ambulatory care for surveillance of an enlarged posterior fontanel. She was born full-term after an uneventful pregnancy and normal vaginal delivery. Physical examination revealed a non-dysmorphic female with normal neurologic examination, a bilateral hip clunk and an open posterior fontanel measuring 1.5 x 1cm. Posterior fontanel was studied at first in neonatal period because of its large diameter and after that due to its persistence. Calcium/phosphorus metabolism and thyroid function were normal. Transfontanellar ultrasound showed a periventricular cyst and hip ultrasound evidenced bilateral developmental dysplasia that required surgery. At 8 months old, the patient was referred to a Neurosurgery center and a cranial CT-scan demonstrated a bilobed bone defect suggesting enlarged parietal foramina, as ilustrated in Figures 1 and 2. There was no relevant family history except for the fact that the patient's father had scaphocephaly, without comorbidities. Watchful

***Correspondence:**
Mariana Gaspar
E-mail: marianagaspar1343@gmail.com

Received: 10-02-2022
Accepted: 23-01-2023
https://pjp.spp.pt

Available online: 01-10-2023
Port J Pediatr. 2023;54(4):275-277
DOI: 10.24875/PJP.M23000051

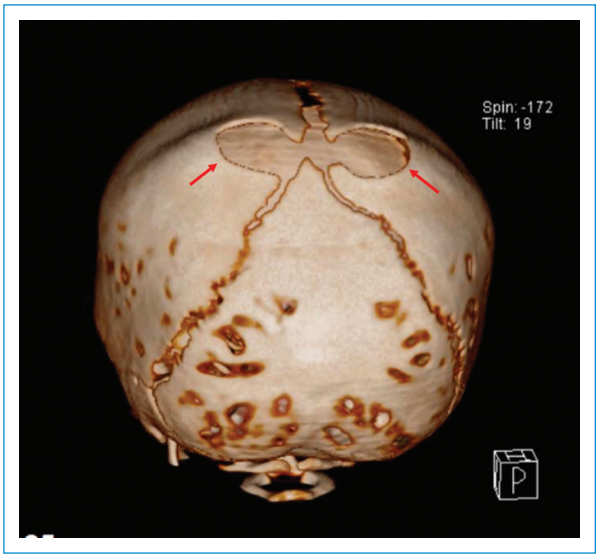

Figure 1. Cranial CT-scan with 3D reconstruction demonstrating a bilobed bone (red arrows).

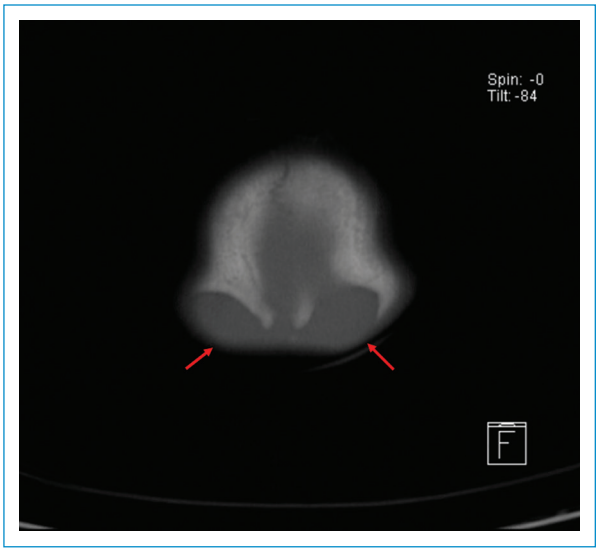

Figure 2. Bone window image of a cranial CT-scan demonstrating a bilobed bone (red arrows).

waiting was recommended with periodic follow-up and parental advice concerning trauma prevention. Genetic testing was not performed. She was followed by both Pediatrics and Neurosurgery until the age of 3, maintaining daily annual follow-up by Neurosurgery. She is now 6 years old and has always remained asymptomatic with normal neurodevelopment and growth including head circumference. Enlarged parietal foramina is a rare, autosomal dominant defect with incomplete penetrance, with a prevalence of 1:15.000-1:50.000[1]. It results from late or incomplete ossification[1,2] ending as large foramina in the upper posterior angle of parietal bone close to the intersection of sagittal and lambdoid sutures[1]. Typically oval or round, enlarged parietal foramina resemble a "pair of spectacles" on postero-anterior skull radiographs and 3D CT scanning using bone windows clearly reveals the defect[3]. Mutations of MSX2 (in 5q34-35) or ALX4 (in 11p11) genes are associated[1,3]. Usually, affected children are asymptomatic but may present with severe headache, vomiting or predisposition for epilepsy[1,3]. It also can be seen associated with anomalies such as myelomeningocele, cleft palate, vascular malformations of the posterior fossa[1] and abnormal venous drainage[2]. Differential diagnosis includes Potocki-Shaffer syndrome, ALX4-related frontonasal dysplasia, MSX2-related craniosynostosis, distal 5q deletions, all including intellectual disability, growth and developmental delay[1]. Apart from genetic disorders, enlarged parietal foramina appears in fetal methotrexate syndrome[1]. This case aims to raise awareness of a rare clinical entity that can be manifested by a persistently opened posterior fontanelle. The prognosis is mostly benign with cranioplasty reserved for very active children, due to risk of trauma, or in cases with associated co-morbidities, such as epilepsy[1].

Awards and previous presentations

The case report was not presented before the submission of the manuscript and prizes were not awarded.

Funding

None.

Conflicts of interest

None.

Ethical disclosures

Protection of human and animal subjects. The authors declare that the procedures followed were in accordance with the regulations of the relevant clinical research ethics committee and with those of the Code of Ethics of the World Medical Association (Declaration of Helsinki).

Confidentiality of data. The authors declare that they have followed the protocols of their work center on the publication of patient data.

Right to privacy and informed consent. The authors have obtained the written informed consent of the patients

or subjects mentioned in the article. The corresponding author is in possession of this document.

Use of artificial intelligence for generating text. The authors declare that they have not used any type of generative artificial intelligence for the writing of this manuscript, nor for the creation of images, graphics, tables, or their corresponding captions.

References

1. Griessenauer CJ, Veith P, Mortazavi MM, Stewart C, Grochowsky A, Loukas M, Shane Tubbs R. Enlarged parietal foramina: a review of genetics, prognosis, radiology and treatment. Child's Nervous System 2013;29:543–7.
2. Fink AM, Maixner W. Enlarged Parietal Foramina: MR Imaging Features in the Fetus and Neonate. American Journal Neuroradiology. 2006;27:1379–81.
3. Mavrogiannis LA, Wilkie AOM. Enlarged Parietal Foramina Synonym: Symmetric Parietal Foramina, U.S. National Library of Medicine, National Center for Biotechnology Information, Gene Reviews, Created: March 30, 2004; Updated: November 27, 2019.