Residência RP Pediátrica
Publicação Oficial da Sociedade Brasileira de Pediatria
ISSN-Online: 2236-6814

Submitted on: 05/05/2019
Approved on: 05/21/2019

CASE REPORT

# Infant macrocephaly: not always an ominous sign

Sângela Fernandes da Silva[1], Lara Samanta Barbosa Ribeiro[1], Nara Lívia Rezende Soares[2], Giuliano da Paz Oliveira[3,4]

**Keywords:**
Hydrocephalus, Megalencephaly, Cephalometry, Child Development.

## Abstract

Benign external hydrocephalus (BEH) is a classically self-limited condition, characterized by macrocephaly in children associated with radiological findings of increase in volume of the subarachnoid space, which may cause ventricular dilation. We report the case of a 19-month-old infant with macrocephaly, mild neurodevelopmental delay, and typical neuroimaging findings. We stress out the importance of early BEH diagnosis in pediatric clinical practice, emphasizing that macrocephaly is not necessarily an ominous signal.

[1] Piauí State University, Medical Student - Teresina - Piauí - Brazil.
[2] Santa Casa de Misericórdia of São Paulo, Pediatric Endocrinology Resident, Pediatrics Department - São Paulo - São Paulo - Brasil.
[3] Federal University of São Paulo, Neuroscience Graduate Program, Neurology and Neurosurgery Department - São Paulo - São Paulo - Brazil.
[4] Federal University of Piauí, Professor at the Medical School - Parnaíba - Piauí - Brazil.

**Correspondence to:**
Giuliano da Paz Oliveira.
Universidade Federal de São Paulo. R. Sena Madureira, nº 1500, Vila Clementino. São Paulo- SP. Brazil. CEP: 04021-001. E-mail: giulianopoliveira@gmail.com

Residência Pediátrica; 2021: Ahead of Print.
DOI: 10.25060/residpediatr-2021.v11n2-133

This work is licensed under a Creative Commons Attribution 4.0 International License.

## INTRODUCTION

Benign external hydrocephalus (BHE), also known as benign subdural effusion, benign subdural collection or benign enlargement of the subarachnoid space, is characterized by increased head circumference (HC) combined with typical neuroimaging findings[1,2]. It has an incidence of around 0.4 per thousand live births, with a higher prevalence in males[1].

The word "benign" reflects the classic view of conceptualizing this condition as self-limiting and paucisymptomatic that occurs in infants with spontaneous resolution during preschool and school age[2,3]. Patients are usually asymptomatic, however the clinical picture may eventually include: dilated scalp veins, mild hypotonia, mild neuropsychomotor developmental delay and intellectual disability[2,4-6]. In some cases, the delay in motor development decreases and disappears within the first four years of life[2]. Macrocephaly (by definition, an increase in head circumference above two standard deviations from the mean) can be present at birth or appear during the first years of life, especially in the first year[1,2].

Cranial neuroimaging findings, whether transfontanellar ultrasound (TFUS), computed tomography (CT) or magnetic resonance imaging (MRI) of the skull, include: enlargement of the subarachnoid space (especially frontal), increased interhemispheric distance and normal or slightly enlarged ventricles[7,8].

HEB is an uncommon disease and little known by pediatricians. The objective of this case report is to describe the importance of the pediatrician's recognition of BHE, as well as to know its clinical manifestations, emphasizing that macrocephaly is not always an ominous clinical sign.

## CASE REPORT

A female infant aged 19 months was brought to the pediatric neurology ward after an enlarged head circumference was observed during a visit to the pediatrician when she was aged 18 months.

The child lives in a rural area in the State of Piauí, in a town called Luís Correia. Her skin is brown, she was born vaginally at 38 weeks of gestation showing good vitality signs, with 1-minute and 5-minute Apgar scores of 8 and 10, and weighing 3.1kg. Her length and head circumference at birth were 49cm and 35.5cm, respectively. Her 24-year-old mother had had two pregnancies before and no miscarriages. She had been to four prenatal appointments and had negative serology for congenital infections.

The infant had mild neurodevelopmental delay. She acquired head control at the age of six months, was able to sit with support when she was eight months old and unsupported at the age of 12 months, waived goodbye when she was 16 months old, and learned her first words ("mama" and "no") and how to walk by herself at the age of 18 months.

Although a pediatrician had been following her at a Basic Healthcare Clinic, measurements of her head circumference had been recorded only twice: 35.5 cm at birth (P95-P97) and 48cm (>P97) when she was 12 months old. Except for macrocephaly evidenced by a head circumference of 50.5cm (>P97, Figure 1), her neurologic findings were normal. She had no tegumental alterations. Her fontanelles were closed, muscle tone was adequate, and tendon reflexes were normal. A skull MRI showed enlargement of the frontal subarachnoid spaces, and a broad interhemispheric fissure with normal ventricles consistent with idiopathic external hydrocephalus (Figure 2).

## DISCUSSION

Macrocephaly is defined as a head circumference above two standard deviations for an individual's age, sex, and body size based on standardized growth charts. It is one of the main causes of referral to a pediatric neurology service. Underlying causes include hydrocephalus (buildup of fluid inside the skull), megalencephaly (abnormally large brain), increased skullcap thickness, and brain hemorrhage[8,9].

Enlarged head circumference may be connected to potentially severe conditions such as BEH, as seen in our patient. Therefore, macrocephaly should not always be perceived as an ominous sign in clinical practice. Some authors found that the presence of developmental delay and focal neurologic signs are relevant in the assessment of children with macrocephaly[9].

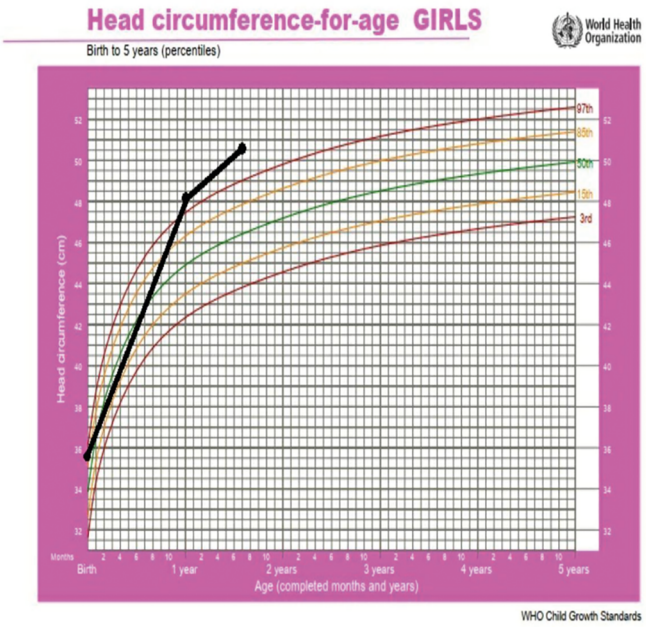



**Figure 1.** World Health Organization's standardized head circumference growth chart.

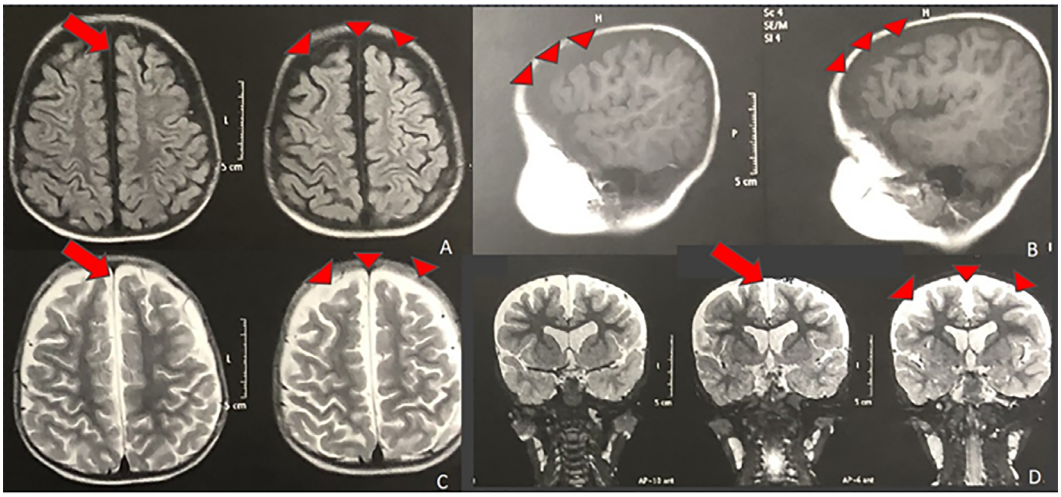



**Figure 2.** Magnetic resonance images of the brain, axial FLAIR (A), sagittal T1 (B), axial T2 (C), coronal T2 (D), showing enlargement of the frontal subarachnoid spaces (arrowhead), a broad interhemispheric fissure (arrow) with normal lateral ventricles.

It is important to note that macrocephaly secondary to BEH may be present from birth or develop later on, particularly during the first year of life. Neonates without macrocephaly that develop BEH usually have a head circumference above the 90$^{th}$ percentile within the first months of life, which in itself is an important clinical sign in the identification of the condition[1,2].

A study carried out in Norway reported that infants were on average younger (mean age of 7.3 months) than our patient at the first appointment with a specialist physician. This may be explained by the comparatively lower levels of access to healthcare in Brazil[1]. Although classically described as an asymptomatic condition, 17.6% of the patients with BEH may present with mild neurodevelopmental delay, as the patient described in this article. Other signs and symptoms that may occur less frequently include dilated scalp veins, hypotonia, vomiting, irritability, epilepsy, and intellectual disability[1,2,6,10,11]. It has been postulated that the pathophysiology of BEH involves an imbalance between the production of cerebrospinal fluid (CSF) in the choroid plexus and CSF resorption in the arachnoid granulations, leading to the accumulation of fluid in the subarachnoid space[12].

Support workup includes transfontanellar ultrasound and head CT and MRI. In our patient, frontal subarachnoid space enlargement and a broad interhemispheric fissure viewed on MRI corroborated a diagnosis of idiopathic benign external hydrocephalus and were in agreement with the literature[1,7,8,10].

Pediatricians must be aware of the causes of macrocephaly, one of which is BEH. Macrocephaly should not always be seen as an ominous sign, particularly in the absence of risk factors such as neurodevelopmental delay and focal neurologic signs. Additional studies are needed to further the knowledge about this condition and its pathophysiology, along with long-term patient follow-up data and prognostic factors.

## REFERENCES

1. Wiig US, Zahl SM, Egge A, Helseth E, Wester K. Epidemiology of benign external hydrocephalus in norway-a population-based study. Pediatr Neurol. 2017 Ago;73:36-41.
2. Zahl SM, Egge A, Helseth E, Wester K. Benign external hydrocephalus: a review, with emphasis on management. Neurosurg Rev. 2011 Out;34(4):417-32.
3. Rekate HL, Blitz AM. Hydrocephalus in children. Handb Clin Neurol. 2016;136:1261-73.
4. Chazal J, Tanguy A, Irthum B, Janny P, Vanneuville G. Dilatation of the subarachnoid pericerebral space and absorption of cerebrospinal fluid in the infant. Anat Clin. 1985;7(1):61-6.
5. Cundall DB, Lamb JT, Roussounis SH. Identical twins with idiopathic external hydrocephalus. Dev Med Child Neurol. 1989 Out;31(5):678-81.
6. Zahl SM, Egge A, Helseth E, Skarbo AB, Wester K. Quality of life and physician-reported developmental, cognitive, and social problems in children with benign external hydrocephalus-long-term follow-up. Childs Nerv Syst. 2019 Fev;35(2):245-50.
7. Gupta SN, Belay B. Intracranial incidental findings on brain MR images in a pediatric neurology practice: a retrospective study. J Neurol Sci. 2008 Jan;264(1-2):34-7.
8. Khosroshahi N, Nikkhah A. Benign enlargement of subarachnoid space in infancy: "a review with emphasis on diagnostic work-up". Iran J Child Neurol. 2018;12(4):7-15.
9. Sampson MA, Berg AD, Huber JN, Olgun G. Necessity of intracranial imaging in infants and children with macrocephaly. Pediatr Neurol. 2019 Abr;93:21-6.
10. Halevy A, Cohen R, Viner I, Diamond G, Shuper A. Development of infants with idiopathic external hydrocephalus. J Child Neurol. 2015 Jul;30(8):1044-7.

11. Mikkelsen R, Rodevand LN, Wiig US, Zahl SM, Bernstsen T, Skarbo AB, et al. Neurocognitive and psychosocial function in children with benign external hydrocephalus (BEH)-a long-term follow-up study. Childs Nerv Syst. 2017 Jan;33(1):91-9.

12. Bradley Junior WG, Bahl G, Alksne JF. Idiopathic normal pressure hydrocephalus may be a “two hit” disease: benign external hydrocephalus in infancy followed by deep white matter ischemia in late adulthood. J Magn Reson Imaging. 2006 Out;24(4):747-55.