Giant cerebellar cavernous malformation in 4-month-old boy. Case report and review of the literature

Olbrzymi naczyniak jamisty móżdżku u 4-miesięcznego niemowlęcia. Opis przypadku i przegląd piśmiennictwa

Elżbieta Jurkiewicz[1], Beata Marcinska[1], Katarzyna Malczyk[1], Wiesława Grajkowska[2], Paweł Daszkiewicz[3], Marcin Roszkowski[3]

[1]Department of Diagnostic Imaging, The Children's Memorial Health Institute, Warsaw, Poland
[2]Department of Pathology, The Children's Memorial Health Institute, Warsaw, Poland
[3]Department of Neurosurgery, The Children's Memorial Health Institute, Warsaw, Poland

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Abstract

Cavernous malformations (CMs) are rare vascular lesions that affect 0.4-0.9% of the population. The diagnosis of CMs is simple in most typical cases although some lesions may present unusual imaging features: localization, signal intensity, or size. Extremely rare giant CMs can mimic neoplastic lesion because of their size. We report a case of giant cerebellar CM that is more than 6 cm in size, diagnosed in 4-month-old boy. We discuss magnetic resonance findings and histopathological features of this lesion.

Key words: magnetic resonance imaging, cerebellum, children, giant cerebellar cavernous malformation.

Streszczenie

Naczyniaki jamiste to rzadkie zmiany naczyniowe, występujące u 0,4–0,9% populacji. Ich rozpoznanie w większości typowych przypadków jest łatwe, ale niektóre zmiany mogą stwarzać trudności diagnostyczne związane z nietypową lokalizacją, ich intensywnością sygnału lub wielkością. Niezwykle rzadkie gigantyczne naczyniaki jamiste mogą imitować zmiany nowotworowe. Autorzy przedstawiają 4-miesięczne niemowlę, u którego rozpoznano olbrzymi naczyniak jamisty zlokalizowany w móżdżku o wymiarze większym niż 6 cm. Omawiają cechy naczyniaka w badaniu za pomocą rezonansu magnetycznego oraz wynik badania histopatologicznego.

Słowa kluczowe: rezonans magnetyczny, móżdżek, dzieci, olbrzymi naczyniak jamisty móżdżku.

Introduction

Cavernous malformations (CMs) are vascular lesions characterized by the presence of multiple dilated vessels with a very slow blood flow. The following synonyms are used in the literature: cavernoma, cavernous angio - ma, cavernous haemangioma, and cerebral cavernous malformation.

Cavernous malformations are easily diagnosed by magnetic resonance imaging (MRI) with reported incidences ranging from 0.4% to 0.9% [1,2]. They comprise 5-13% of intracranial vascular anomalies [3]. Cerebral CMs are rarely described in children. The diagnosis of CMs is simple in most typical cases, although some lesions may present unusual imaging features such as localization, signal intensity, or size. The majority of CMs

Correspondence address: Elżbieta Jurkiewicz, Zakład Diagnostyki Obrazowej, Pracownia Rezonansu Magnetycznego, Instytut „Pomnik – Centrum Zdrowia Dziecka", Al. Dzieci Polskich 20, 04-730 Warszawa, fax: +22 815 12 76, e-mail: e-jurkiewicz@o2.pl
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are small, but they may reach a significant size. Giant CMs are exceptional in paediatric patients and can mimic a neoplastic lesion. Magnetic resonance imaging with gradient- or susceptibility-weighted sequences is the method of choice in diagnostics. Cavernous malformations may be seen as an incidental finding on MRI studies, or they may present with symptoms. Recurrent haemorrhage and haemosiderin deposits are often observed.

In this paper, we report a case of giant cerebellar CM more than 6 cm in size that was diagnosed in a 4-month-old boy. We discuss the MRI findings and histological features of this lesion. We report this case because of the rarity of a giant cerebellar CM in children in the first year of life.

Case report

A 4-month-old boy was admitted due to intracranial hypertension syndrome and anxiety attacks that appeared one week before admission to the hospital. These symptoms occurred only in the day and lasted up to 30 minutes. On examination he was found to have a tense anterior fontanellae and setting sun sign. His head circumference was significantly increased compared with previous measurements.

Computed tomography (CT) of the brain performed on admission showed a huge mass located in the right hemisphere of the cerebellum with areas of bleeding. The supratentorial ventricular system was markedly dilated and signs of obstructive hydrocephalus were present (Fig. 1). Treatment was begun by urgent implantation of a high-pressure ventriculo-peritoneal shunt. This modality was chosen because of the need to relieve hydrocephalus while preventing upward incisural herniation.

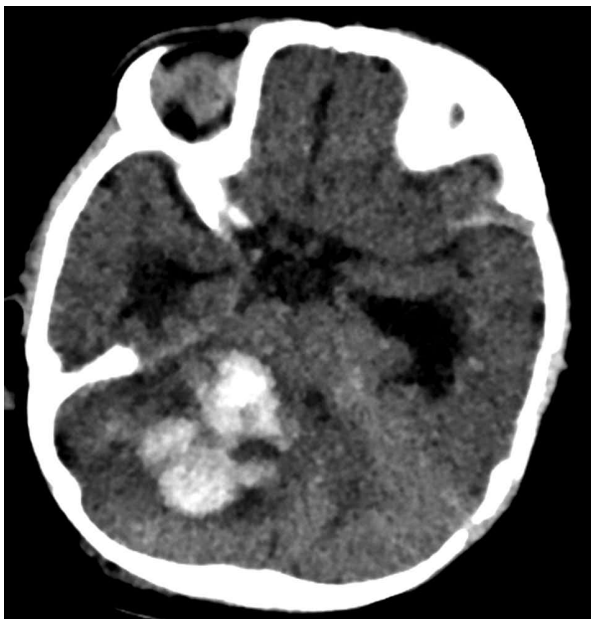

Fig. 1. CT scan at the time of admission. There is a hyperdense haemorrhagic lesion in the right cerebellar hemisphere. Widening of the temporal horns of lateral ventricles is seen

Brain MRI was subsequently performed on a 1.5 T scanner. Magnetic resonance images showed a huge multicystic lesion of 59 × 45 × 61 mm, located in the right cerebellar hemisphere causing midline shift and obstructive hydrocephalus. Compression of the fourth ventricle was seen. The lesion had well-defined borders, showed mixed intensities, with areas of hyperintensity representing haemorrhage, surrounded by a low-signal-intensity, thin rim representing hemosiderin (Figs. 2A-D). Cerebellar CM was suggested. The main differential diagnosis included atypical vascular malformation and haemorrhagic neoplasm.

The gradient sequence and susceptibility weighted images (SWI) additionally revealed numerous small, low-signal lesions corresponding to multiple small cavernomas located in both cerebral hemispheres and in the left cerebellar hemisphere; some of them are presented on an axial SWI image (Fig. 3).

The patient was operated on with right suboccipital craniotomy. The well-capsulated and multiseptated lesion was resected.

Microscopically, the lesion was composed of a mass of dilated, thin-walled, vascular sinusoids with little intervening brain tissue (Figs. 4A-B). The peripheral cerebellar parenchyma revealed the presence of numerous haemosiderin-laden macrophages, gliosis, and foci of mineralization. The diagnosis of cerebellar CM was established. Histologically, these lesions are considered vascular malformations rather than true tumors called cavernous haemangioma. Historically, cerebellar CMs have been defined as having no intervening brain tissue. Recent studies have shown, however, that such tissue can occur between abnormal vascular channels containing only a single layer of endothelium without a muscular layer or internal elastic lamina.

The patient made a good postoperative recovery with improvement in his neurological status. A follow-up CT examination performed 7 weeks after surgery showed residual cavernoma within the right cerebellar hemisphere (Fig. 5). The patient's parents declined to continue further treatment at our Institute.

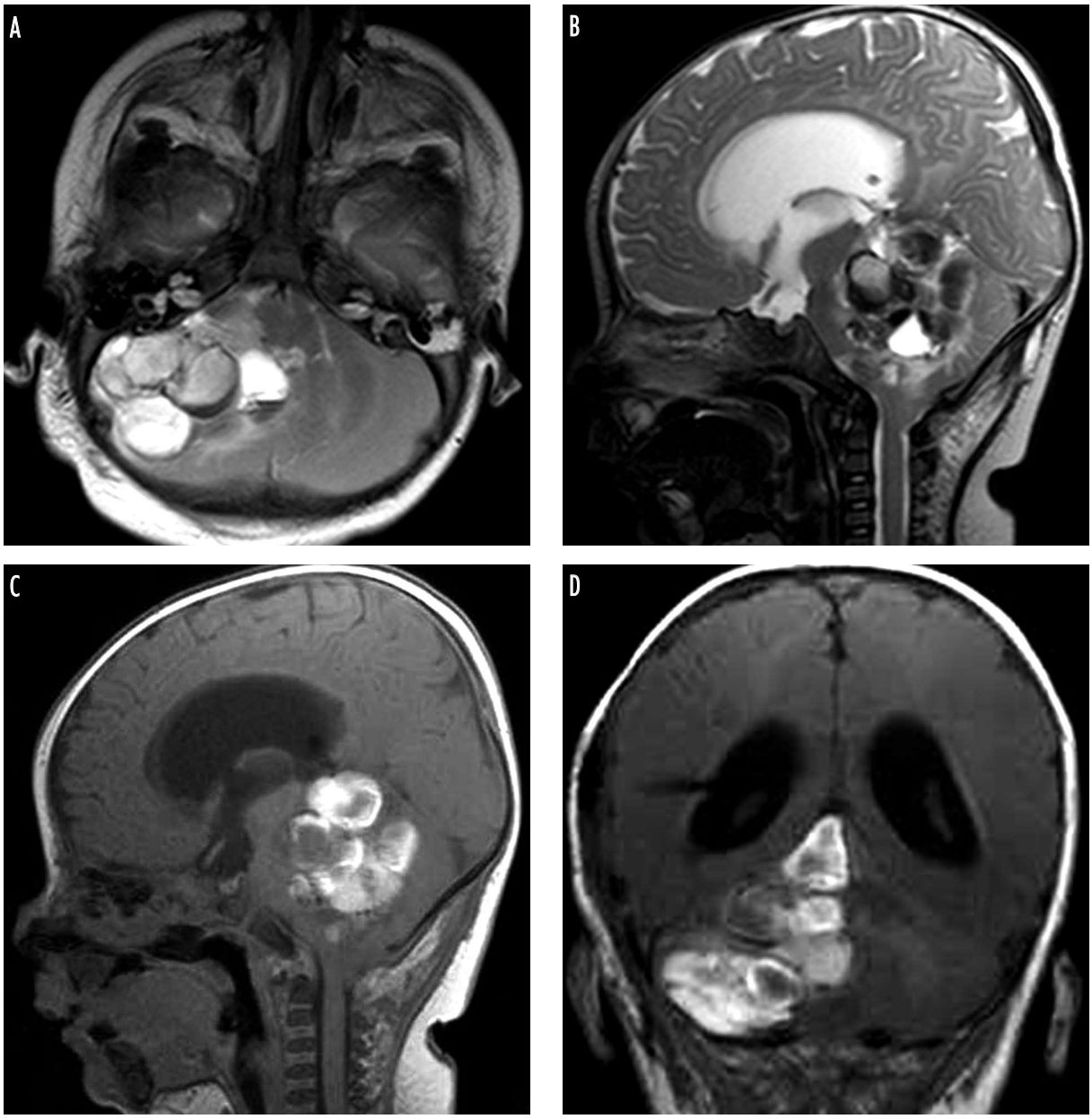

Fig. 2. MRI: T2-weighted axial (A) and sagittal (B) images show giant cerebellar cavernous malformation located in the right cerebellar hemisphere with mixed intensity of nodules and fluid-fluid levels within the lesion. The periphery of the mass shows a low intensity. T1-weighted sagittal (C) and coronal (D) images demonstrate multicystic huge cavernous malformation with high signal indicative of haemorrhage. Cerebellar tonsils are displaced down into foramen magnum. Reduction of the prepontine cerebrospinal space is visible

Discussion

Cavernous malformations represent 5-13% of intracranial vascular anomalies. They are most commonly located supratentorially, particularly in the parietal lobe and thalamus. Infratentorial CMs are found in the brain stem and, sporadically, in the cerebellum or in the spinal cord [4-8]. Cerebral CMs are rarely described in children.

Cavernomas vary in size from a few millimeters to a few centimeters, with the majority of lesions being small. Giant CMs are very rare and occur predominantly as solitary lesions in the supratentorial compartment. The definition of giant CMs is arbitrary, however. Lawton *et al*. defined a giant cavernous haemangioma as a cavernoma exceeding 6 cm in at least one dimension, but in accordance with Kan *et al*., cavernomas with

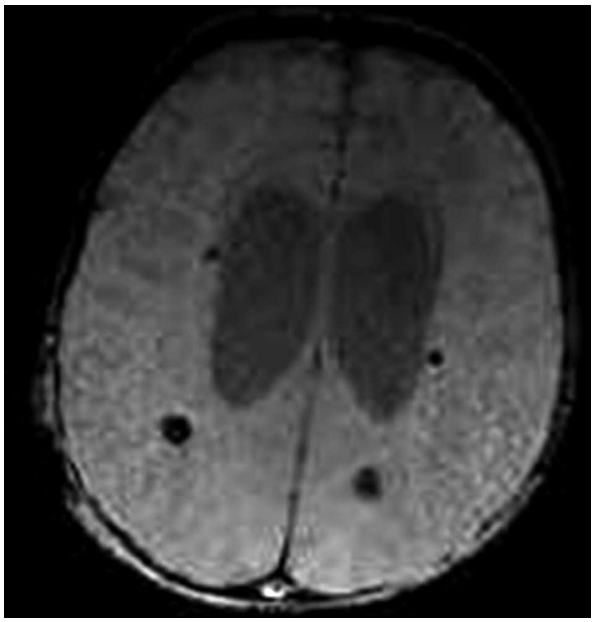

Fig. 3. SWI sequence shows some of the multiple hypointense lesions in the both cerebral hemispheres

a diameter greater than 4 cm are defined as giant [2,9]. Our patient fulfilled Lawton's definition of size with one diameter of more than 6 cm.

Posterior fossa giant CMs are extremely rare. According to our knowledge, only 13 cases of giant CMs have been reported in this localization in the English-language literature [2,4-7,10-12]. Nine of them were located in the cerebellar parenchyma [2,4,6,7,11,13]. Among the published cases, there are only 4 of giant lesions with a diameter more than 6 cm (of which three were located in the cerebellum) as described by Lawton *et al*. in 2004 [9]. Our case of a 4-month-old infant is the fourth cerebellar giant CM. The data of previously published posterior fossa cases are presented in Table 1.

Giant CM may present a varied image in MRI and CT studies. On CT examination, typical lesions are well-circumscribed, frequently hyperdense, and have cystic components, calcifications, and areas of haemorrhage. The cystic type of giant cavernoma is rarely found [3].

Magnetic resonance imaging shows well-delineated hyperintense lesions, with a typical 'salt and pepper' appearance on T1-weighted images. A mixed, heterogenous signal seen on T2-weighted images represents haemorrhage in different stages of evolution. A low hemosiderin signal typically surrounds the lesion. Gradient echo sequence and SWI are the most sensitive for imaging hemosiderin because of the magnetic susceptibility effect. There is no enhancement in the majority of cases, although contrast enhancement has sometimes been seen. It can be present in delayed images or in dural cavernoma, in particular [1].

Oedema and mass effect on the surrounding brain or adjacent ventricular system can be present. Because of the variable appearance of CMs, posterior fossa neoplasms, metastases, and other vascular malformations should be considered in the differential diagnosis, especially in cases with significant haemorrhage, mass effect, and oedema.

Small cavernomas are frequently asymptomatic and are detected serendipitously. They do not require surgery, whereas giant CMs present varied symptoms related to their supra- or infratentorial localization. In a recently published paper, the authors reported a population-based study of 139 adults, with intracranial haemorrhage diagnosed in 17 patients (12%). The risk was higher in lesions located in the brainstem: 32% vs. 5% with other

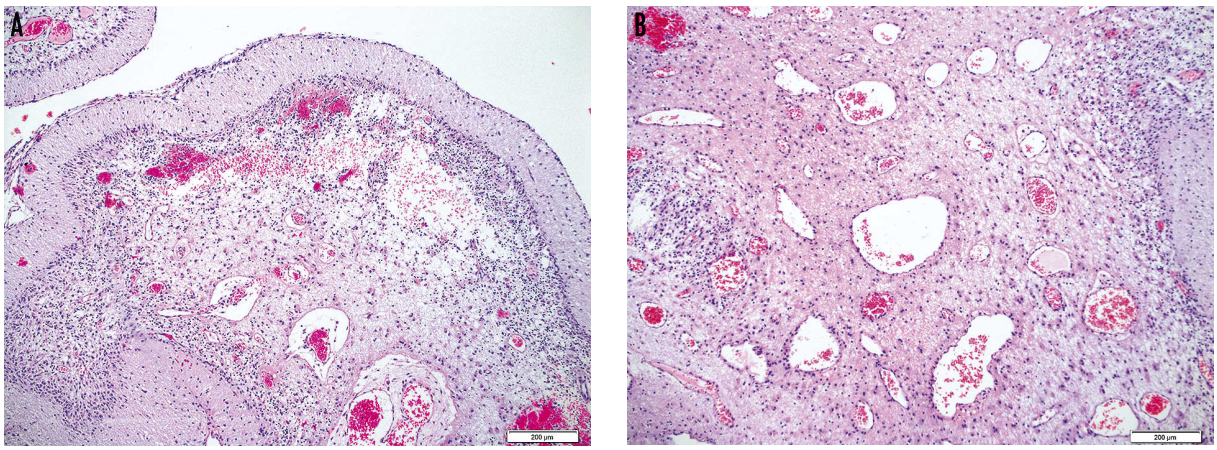

Fig. 4. Histopathology: Cerebellar cavernous malformation. (A) Cerebellar cortex with the presence of dilated, thin-walled vascular sinusoids. H&E staining. (B) Thin-walled vascular sinusoids surrounded by gliosis. H&E staining

localizations. The authors concluded that the 5-year risk of the first intracranial haemorrhage was lower than the risk of recurrence (2.4% vs. 29.5%) [14]. Because of the small number of published cases, there is nostatistical data related to the incidence of bleeding of congenital giant CMs located in the cerebellum, but haemorrhages in this localization are life-threatening. Giant lesions located in the cerebellum more likely lead to obstructive hydrocephalus, as in our case. On admission, the child demonstrated signs of rapidly increasing hydrocephalus with signs of progressive herniation. The CT scan suggested haemorrhage from a vascular malformation. For this reason, it was decided to first relieve the symptoms of obstructive hydrocephalus to enable subsequent MRI diagnostics. Surgery was then performed because this was judged to be the best modality. According to the surgical report, the lesion was radically resected. Nonetheless, experience at our institution shows that the frequency of discrepancy between postoperative MRI/CT imaging and the surgical report is about 15-20% in the case of cavernous angiomas located in the posterior fossa.

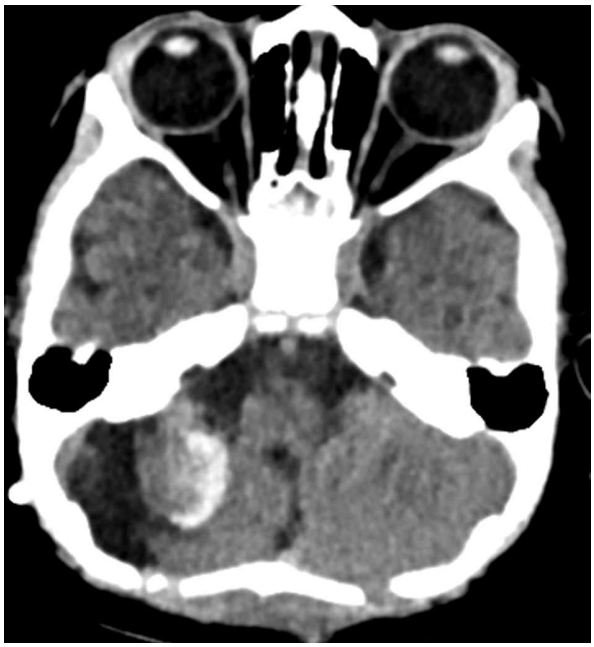

Fig. 5. Follow-up CT scan showed residual cavernoma within right cerebellar hemisphere

Conclusions

Cerebellar CMs, whether solitary or multiple, are uncommon lesions, that can reach a very large size and cause bleeding. Giant cerebellar CMs in children in the first year of life are extremely rare. The presence of hemosiderin products should lead to consideration of giant CM, and MRI allows differentiation between cavernomas and neoplasms.

Table 1. Giant posterior fossa cavernous malformations reported in the literature

No	Author, year [reference]	Age	Sex	Size, cm	Localization
1	Greene, 1986 [6]	2 months	M	nd	Cerebellar hemisphere and vermis
2	Kan, 2008 [2]	25 years	F	4.0 × 3.0 × 3.0	Left cerebellum paramedian
3	Kan, 2008 [2]	2 months	M	4.0 × 3.0	Cerebellum paramedian
4	Kan, 2008 [2]	nd	nd	5.0 × 4.0	Left cerebellar hemisphere extending to the left pons
5	Thiex, 2003 [9]	26 years	F	nd	Brainstem
6	Yasui, 2005 [10]	42 years	F	nd	Brainstem
7	Tripathy, 2009 [11]	46 years	M	4.2	Left cerebellar hemisphere
8	Lew, 2010 [12]	4 months	M	4.0	Cerebellum
9	Lew, 2010 [12]	7 months	F	nd	Right cerebellar hemisphere extending to the pons
10	Hayashi, 1985 [5]	6 months	F	6.0	Both cerebellar hemispheres and vermis
11	Braga, 2004 [7]	9 months	F	5.3 × 3.8 × 6.1	Brainstem
12	Atalar, 2007 [4]	6 years	F	4.0 × 3.5 × 6.0	Left cerebellar hemisphere
13	Kan, 2008 [2]	1 month	M	6.0 × 6.0	Cerebellum
14	Present study	4 months	M	5.9 × 4.5 × 6.1	Right cerebellar hemisphere

M – male, F – female, nd – no data

Disclosure

Authors report no conflict of interest.

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