

Cavernous Angioma Presenting as Encephalocele: Two Case Reports and Review of the Literature

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Abstract: A cavernous angioma (also known as a cavernous malformation or cavernoma) is a type of vascular malformation. Consisting of a low-pressure collection of vein-like vessels in the shape of caverns, cavernous angioma can occur anywhere in the brain or spinal cord. While it was originally believed that most vascular malformations are present at birth (congenital), cavernous malformation lesions may develop throughout the lifetime of the affected individual. There are no documented cases of cavernous malformation being the lead point in encephaloceles in current literature to our knowledge. Cavernous malformation presents as headaches, progressive neurologic deficits, seizures or bleeding but not as encephaloceles. We report two neonates, a male and female with histopathologic diagnosis of cavernous angioma that presented as frontonasal and occipital encephalocele.

Key words: Cavernous angioma, vascular malformation, neonates.

1. Introduction

Vascular malformations are localized collections of blood vessels that are abnormal in structure or number, lead to altered blood flow, and are not cancerous [1]. While it was originally believed that most vascular malformations are present at birth (congenital), cavernous malformation lesions may develop throughout the lifetime of the affected individual. Other vascular malformations are not congenital, but are caused by trauma, radiation, or other injury to the spinal cord [1, 2].

Cavernous malformations are dilated blood vessels that are characterized by multiple distended “caverns” of blood-filled vasculature through which the blood flows very slowly [1-3]. Vessels of a cavernous malformation lesion have a tendency to leak because they lack the proper junctions between neighboring cells as well as the necessary structural support from smooth muscle and stretchable material [4]. Leakage

(bleeding) from these vascular lesions is the underlying cause of clinical symptoms associated with the illness [5].

Cavernous malformations are primarily located in the brain, but can also be found in the spinal cord, on the skin, and more rarely in the retina [3, 6].

Cerebral cavernous malformations (CCMs) are usually located in the white matter (cortex) of the brain. CCM do not have brain tissue within the malformation like other lesions such as arteriovenous malformations, and they usually do not have defined borders. CCM are dynamic structures, changing in size and number over time and they can range in size from a few millimeters to several centimeters [3, 6].

CCM are present in up to 0.5% of the general population, and they account for a large proportion (8%-15%) of all brain and spinal vascular malformations. While the prevalence of individuals with at least one CCM lesion is quite high, as many as 40% of affected individual may never experience symptoms or become diagnosed with cavernous malformation [1, 3, 6].

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Majority of these cases are sporadic with only a single lesion and no family history of the disease [7]. Individuals with the familial (genetic) form of cavernous malformation are likely to have multiple lesions and may be more likely to experience symptoms associated with the disorder [7, 8].

While adults are most often diagnosed with CCM, people of all ages may be affected by cavernous malformations, and approximately 25% of all diagnosed cavernous malformations are found in children.

There are no documented cases of cavernous malformation being the lead point in encephaloceles in current literature to our knowledge. Cavernous malformation presents as headaches, progressive neurologic deficits, seizures or bleeding but not as encephaloceles.

We report two neonates, a male and female with histopathologic diagnosis of cavernous angioma that presented as frontonasal and occipital encephalocele.

2. Case 1

A term female neonate delivered to a 32 year-old healthy Nigerian mother, who was gravida 3, para 0, from a non-consanguineous marriage. Gestation was

complicated in the 1st month with high-grade fever managed only with paracetamol. There was unremarkable family history. At birth, the neonate was noted to have ulcerated frontonasal protrusion without cerebrospinal fluid (CSF) leak and a dysmorphic face with associated microcephaly, and pancraniosynostosis. There was hypertelorism and a shallow orbit but no syndactyly. She weighed 2.0 kg, and had an occipitofrontal circumference (OFC) of 27 cm. A cranial CT scan showed features of alobar holoprosencephaly with a monoventricle, absent basal ganglia and corpus callosum, a frontonasal defect with an encephalocele. On the 5th day of life she had excision of the frontonasal encephalocele and repair of the frontal defect.

Histologically, the excised tissue was described as cavernous angioma. Postoperatively, she did well with no new neurological or feeding problems

Microscopic examination shows proliferation of cystically dilated small-sized vascular channels filled with red blood cells causing herniation of the cerebral cortex. The vascular channels borders one of the ventricles. The histopathologic diagnosis was confirmed to be cavernous angioma in a female neonate (Fig. 1).

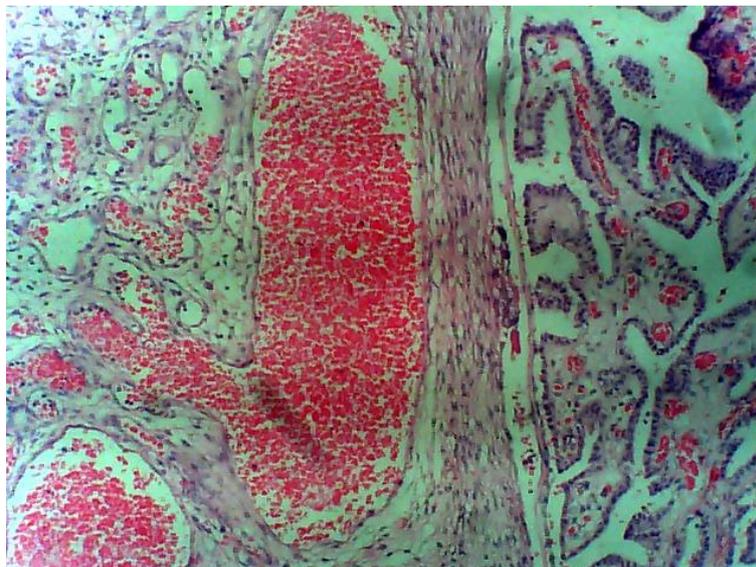


Fig. 1 Cavernous Angioma of the fronto-nasal region causing herniation of the cerebral cortex and part of the choroid plexus. HE $\times 100$.

3. Case 2

A term, male, infant who is the firstborn to a 17-year old single mother and had presented at 3 weeks post-delivery with a skin-covered occipital protrusion from birth. Delivery was normal. There were no symptoms suggestive of other congenital anomalies and there was no family history of any congenital anomalies.

Child's mother ingested traditional and unprescribed medications during the 1st month of gestation, prior to discovery that she was pregnant, for lower abdominal pains.

Examination showed a normal sized head with an infratocular, predominantly cystic occipital swelling with a circumference of 4 cm and length of 8 cm. He had no other dysmorphic features nor other physical abnormalities.

A diagnosis of infratorcular occipital encephalocele was made.

He had excision and repair with an intra-operative finding of distended, protruding arachnoid membrane containing clear CSF and a small tongue of dysplastic brain tissue, which was sent for pathologic examination.

Microscopic examination shows proliferation of cystically dilated small-sized vascular channels filled

with red blood cells causing herniation of the cerebral cortex. The histopathologic diagnosis was confirmed to be cavernous angioma in a male neonate (Fig. 2).

4. Discussion

Vascular malformations in CNS (Central nervous system) are divided into four groups; namely, arteriovenous, venous, capillary telangiectasis and cavernomas [9]. Cavernoma (cavernous hemangiomas, angiomas, cavernous malformation) is a benign vascular lesion occurring at any site within the CNS.

The etiology of CA (Cavernous Angioma) is not known. A small number of cases are hereditary with high penetrance and autosomal dominant transmission. Recently, a gene has been mapped to chromosome 7q11-q22 in Hispanics and some white people, but not in all families [7, 8]. The hereditary form is highly prevalent in Hispanics and North Americans. CA is assumed to be congenital, as they do not display any neoplastic features [10].

A correlation between radiation and cavernomas of the brain has been determined, particularly in case of a positive history of radiation exposure in childhood [11].

Macroscopically, these resemble a ripe mulberry and microscopically they contain blood filled cavities lined

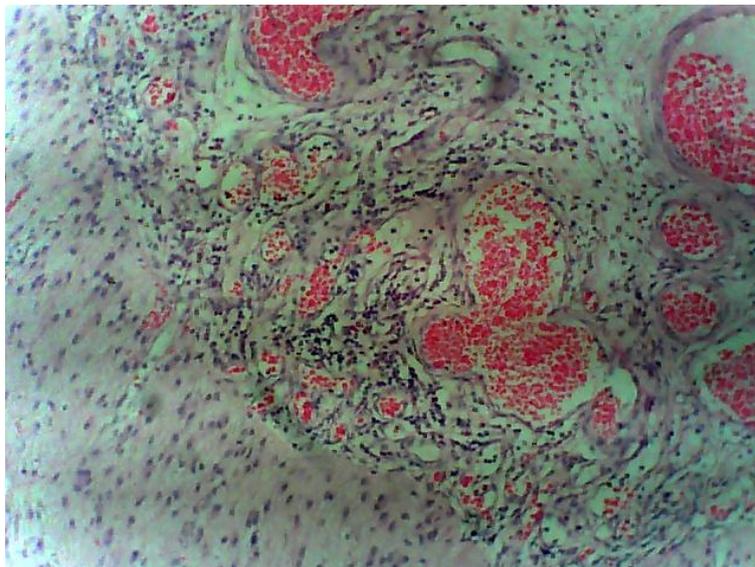


Fig. 2 Cavernous Angioma of the occipital region causing herniation of the cerebral cortex. HE $\times 100$.

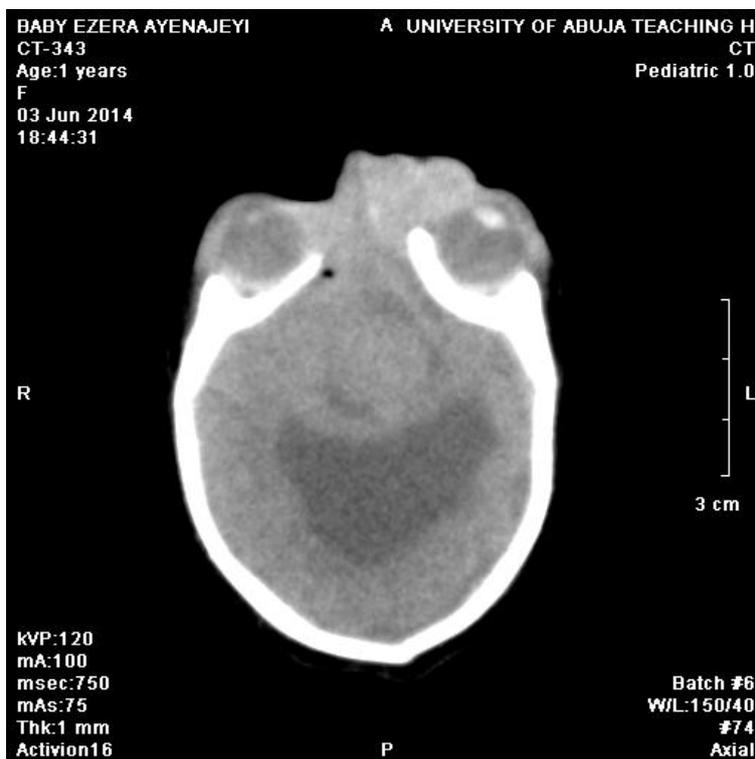


Fig. 3 The CT Scan of the first neonate that presented with frontonasal encephalocele, but was later found to have alobar holoprosencephaly.

by a single layer of endothelium and separated by neuroglia without any neural tissue [12].

CT (Computerized tomography) demonstrates these lesions as focal high attenuation masses with variable calcification in the absence of edema or mass effect. Enhancement is typically mild on CT, which may not be identifiable [13].

MRI (Magnetic resonance Imaging) is reliable in the detection, follow up and diagnosis of symptomatic and asymptomatic CA. CA appear as a reticulated core of mixed signal representing blood in various stages of degeneration surrounded by a hypointense halo due to hemosiderin on T2W MRI [14].

In the female neonate (case 1) that was born with ulcerated frontonasal cerebral herniation, the CT scan done did demonstrate a focal high attenuation mass anterior to the left eyeball but no calcification and also revealed alobar holoprosencephaly (Fig. 3) [15], while the male neonate that was born with occipital encephalocele couldn't afford CT scan due to financial constraint.

However, MRI that is the gold standard for diagnosis couldn't be done due to the high cost of this procedure in our environment. Both neonates could afford surgery and with the histopathology examination diagnosis of cavernous angioma was made in both cases.

CA varies in size from a few millimeters to a few centimeters. The majority of them is small, but may reach a significant size. Those measuring more than 6 cm in diameter are termed as giant CA [16]. The size of the two lesions in this review ranged from 4-8 cm in diameter, thus suggesting that their huge sizes may have interfered with the normal embryology in utero, thereby causing the encephaloceles. Embryologically, it has been known that the area cerebrovasculosa of the telencephalon consists of haphazardly oriented mature and immature neurons, glial cells, and nerve fibres. This neural matrix is perfused by extensive proliferation of small thin-walled vascular channels, so concentrated in places to resemble a cavernous angioma [17].

Patients with CA are often asymptomatic and when

symptoms are present, depend on the location and size of the lesion. Most frequent presentations are focal neurological deficits, hemorrhage and epilepsy [18]. There are no documentations in current literature as to CA presenting as encephaloceles.

Both neonates had good cosmetic outcome at the age of 3 months; however, the female neonate had problems with fixation of eyes on moving objects, social smile and neck controls due to the underlying brain pathology of alobar holoprosencephaly. This is to be expected, as lesions in the fronto-temporal region have behavioural and memory disturbances [14].

MRI findings are diagnostic of CA, which usually do not require any further confirmation. By any means; however, in very rare cases of ambiguity, some of the differential diagnoses which may be thought of are hypothalamic glioma, germinoma, hamartoma, histiocytosis and granuloma but presence of mixed intensity “popcorn” like core and a hypointense hemosiderin rim helps in ruling out these conditions [14].

A CT scan may show an encephalocele but the findings of the contents are not pathognomonic of CA. The findings of an MRI are more pathognomonic with the addition of the T2W gradient echo. The role of pathological examination of specimens is buttressed as this diagnosis would have been impossible.

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