

Focal neurological symptoms associated with cavernous malformation

Introduction

This case reports the atypical presentation of a young woman who suffered headaches associated with a subtle and isolated left fourth cranial nerve palsy. Whereas initial review dismissed her symptoms as ‘migraine’, detailed ophthalmology and imaging assessments revealed that her symptoms occurred following decompensation (thrombosis and intralesional bleed) of a cerebral cavernous malformation. Her symptoms resolved spontaneously over 2 months. This article discusses the unique anatomical features, diagnosis and management of cavernous malformations, with particular consideration of the risks of bleeding and the need for neurosurgical opinion within a multidisciplinary team to decide on the best patient-centred management strategies.

Discussion

Cavernomas (or cavernous haemangiomas) are low flow vascular malformations of the central nervous system. There are both sporadic (80%) and familial forms, with sporadic forms usually presenting with an isolated lesion (Flemming and Lanzino, 2020). Cavernomas are often asymptomatic and are the second most common incidental finding on brain magnetic resonance imaging (Horne et al, 2016). Asymptomatic cavernomas affect 0.2–0.4% of the population (Horne et al, 2016).

Case report

A woman of South Asian ethnicity, in her thirties, presented with a 5-day history of sudden onset headache, diplopia and non-positional dizziness. She had known factor VIII deficiency which had been incidentally diagnosed during a previous pregnancy, and was not taking any medications. She attended her local emergency department twice, and was advised that there were no deficits on examination. She then obtained an urgent specialist ophthalmology opinion, which identified a left 4th nerve palsy, with a compensatory right-sided head tilt. They referred her urgently to the hyperacute stroke unit, where her observations were found to be normal, and no further neurological deficits were observed.

An acute computed tomography scan of the head demonstrated a hyperdense focus in the right medial thalamus and rostral midbrain, which raised concerns of haemorrhage. There was no abnormality on computed tomography cerebral angiogram. Her coagulation profile revealed a prolonged activated partial thromboplastin time of 125 seconds. She was treated with a short course of tranexamic acid, based on advice from haematology.

A few days later during her admission, she underwent more detailed magnetic resonance imaging of her brain, which revealed a small, mixed-signal intensity lesion with internal fluid levels and a haemosiderin rim, in keeping with a cerebral cavernous malformation with intralesional haemorrhage within the right medial thalamus. The inferomedial midbrain component was associated with T1 shortening, diffusion restriction and perilesional oedema (Figure 1). In this case, the patient’s unique deficit of a left superior oblique palsy was caused by the lesion encroaching on the contralateral, right trochlear nucleus. This presentation was therefore a result of intralesional haemorrhage and thrombosis of a cavernous malformation without rupture.

Her case was discussed at the local neurovascular multidisciplinary meeting. Given the patient’s clinical stability, the isolated and unruptured lesion and the surgically high-risk location, it was decided that medical management was most appropriate. She was treated with pain relief. A repeat magnetic resonance imaging 2 months later demonstrated appropriate resolution of the mild intralesional haemorrhage and oedema, and follow-up orthoptic assessment showed parallel resolution of her diplopia.

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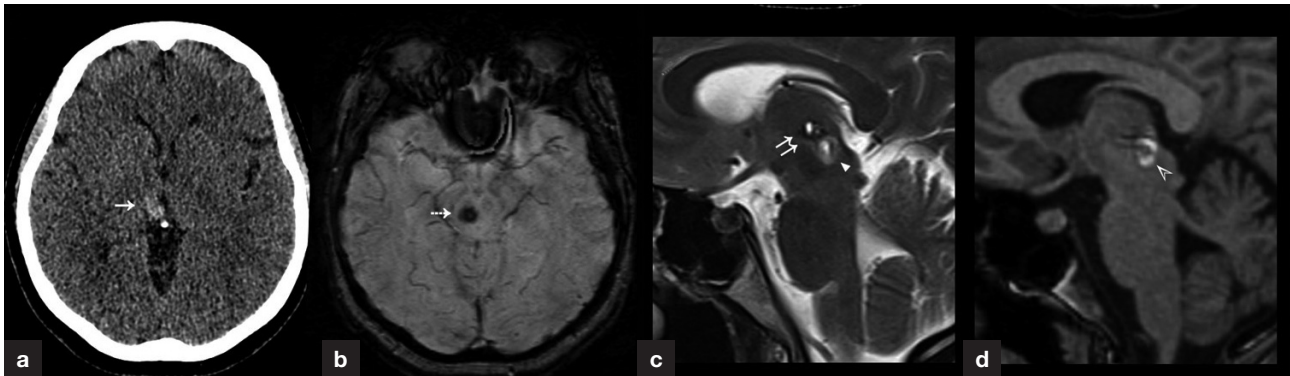


Figure 1. Imaging features of a symptomatic right thalamo-mesencephalic cavernous malformation. a. Axial computed tomography demonstrates a focal area of hyperdensity in the medial right thalamus (single arrow), with mild mass effect on the adjacent third ventricle but no hydrocephalus. On (b) axial susceptibility-weighted, (c) sagittal T2-weighted and (d) sagittal T1-weighted magnetic resonance images, this region corresponds to a bilobed, mixed intensity lesion in the right medial thalamus and midbrain, with blooming hypointensity on susceptibility-weighted imaging (dashed arrow in b) and a haemosiderin rim (double arrows in c). Magnetic resonance imaging is the most sensitive and specific investigation for cavernomas.

When symptomatic, patients most commonly present with focal neurological deficits, seizures and/or acute headache. Neurological deficits, such as diplopia in this case, present as a result of mass effect from thrombosis and/or intralesional haematoma of the cavernoma compressing pertinent structures in the brain. A more severe complication occurs when there is rupture of the cavernoma with blood extending to brain tissue or adjacent CSF. Only 15% of adults with symptomatic cavernomas present with intracranial haemorrhage at the time of diagnosis (Salman et al, 2008). Risk factors for haemorrhage in sporadic cases include a recent history of previous haemorrhage as a first clinical presentation and a lesion located in the brainstem (Salman et al, 2008; Flemming and Lanzino, 2020).

Asymptomatic cavernomas can be managed conservatively. Symptomatic cavernomas may be managed medically or surgically. Clinical trials are ongoing to determine evidence for medical therapies including propranolol, statins and the development of novel drugs targeting biomarker-driven biochemical pathways (Hegde et al, 2012). A paradoxical reduction in haemorrhagic events was reported in a meta-analysis in patients treated with antithrombotics, with the benefit thought to result from the prevention of intralesional thromboses (Horne et al, 2016). If safely targetable, surgical resection or stereotactic radiosurgery may be considered when there is recurrent symptomatic haemorrhage or medically resistant epilepsy (Leksell, 1983; Snellings et al, 2021). The ‘Cavernomas – A Randomised Effectiveness (CARE)’ pilot trial comparing neurosurgery or stereotactic radiosurgery with medical therapy in symptomatic cavernomas (<https://clinicaltrials.gov/ct2/show/NCT02736669>) will be important in providing evidence for the management of cavernomas in the future.

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Learning points

- The peculiarities of the fourth (trochlear) nerve should be kept in mind when performing assessments.
- Cavernomas are one of the most common incidental findings on imaging.
- Clinicians should follow appropriate evidence for managing patients with cavernomas whether they are symptomatic or not, and discuss these cases in multidisciplinary meetings based on their location and bleeding risk.
- Cavernomas have characteristic imaging appearances, the presence of intrinsic high T1-weighted signal and surrounding oedema suggests recent intralesional haemorrhage, likely resulting from a thrombosis.

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