A girl with acute-onset severe astigmatism and gaze palsy

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▸ A video clip demonstrating physical examination findings of bilateral fixed left lateral gaze and right lower-motor-neuron seventh cranial nerve palsy is available at www.hkmj.org

A previously healthy 4-year-old girl presented with a 4-week history of acute-onset visual disturbance. She was initially prescribed glasses for severe astigmatism but vision did not improve. It was later noted by her parents that her eyes could look only to the left side. She was then admitted to hospital. There was no history of headache, photophobia or vomiting but she exhibited drooling from the right side of her mouth and unsteady gait. She was afebrile and all other vital signs were normal with Glasgow Coma Scale score of 15. Physical examination revealed bilateral fixed left lateral gaze and right lower-motor-neuron seventh cranial nerve palsy. When asked to look to her right, she needed to compensate by head turning. Pupils were 3 mm equal and reactive to light. Ear canals were normal and there was no skin rash. Computed tomography scan of the brain showed a multilobulated mixed hyperdense and hypodense lesion at the dorsal pons with compression of the fourth ventricle. Magnetic resonance imaging (MRI) of the brain revealed bleeding from a tumour at the right dorsal pons (Fig 1); overall picture suggested multiple cavernoma (Fig 2). Neurosurgical decompression and stereotactic excision of a brainstem lesion was performed via suboccipital craniotomy. Histology showed features of a vascular lesion in keeping with cavernous haemangioma. Intracranial pressure was monitored via an external ventricular drain and remained normal postoperatively. Postoperative neurological examination showed bilateral fixed left lateral gaze, right lower-motor-neuron seventh

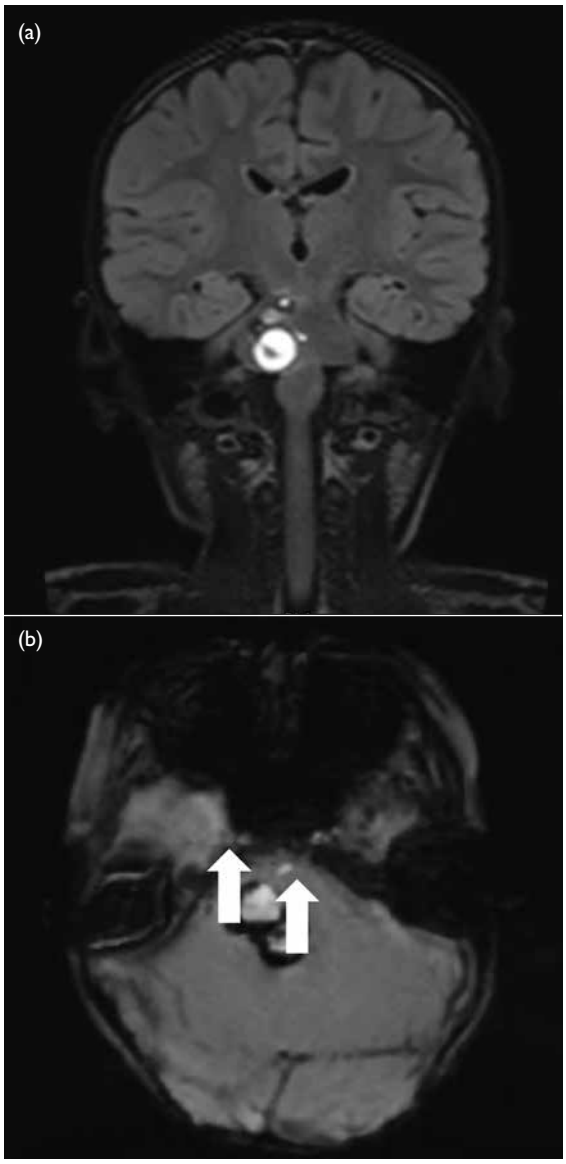

FIG 1. (a) Coronal fluid-attenuated inversion recovery imaging sequence and (b) axial susceptibility weighted imaging sequence showing lobulated T2-hyperintense lesion with bleeding (arrows) at the right dorsal pons

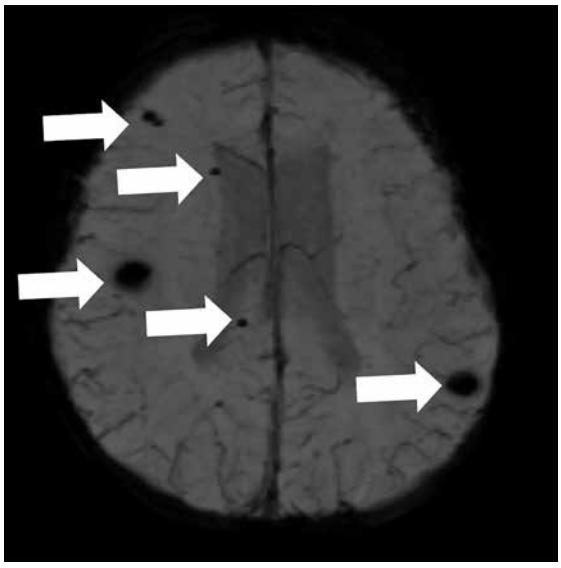

FIG 2. Magnetic resonance imaging axial susceptibility weighted image showing additional foci demonstrating susceptibility artefacts (arrows) in bilateral cerebral hemispheres, compatible with multiple cavernoma

cranial nerve palsy, and contralateral weakness of limbs. Genetic testing demonstrated a pathogenic mutation [heterozygous KRIT1 c.690C>A p.(Tyr230*)] for familial cerebral cavernous malformation (CM) syndrome. Her family was referred for further genetic testing and counselling.

Acute-onset gaze disturbance in children is alarming and may signify serious cerebral abnormality. Almost all conjugate gaze palsies originate from a lesion in the midbrain or pons. These lesions can be caused by vascular or oncological space occupying lesions.

Cavernous malformations, also known as cavernous haemangiomas, are a type of benign, congenital malformation in which a cluster of dilated thin-walled capillaries form a characteristic 'mulberry' lesion with engorged purplish colour.[1] They can be sporadic or inherited with an autosomal dominant pattern and incomplete penetrance, and can present as solitary or multiple lesions. Unlike capillary haemangiomas, CMs can be life-threatening and do not tend to regress. In all, 25% of cerebral CMs are infratentorial. They have a bleeding rate of 2% to 3% per year and recurrent bleeding rate of >20%. Due to the close proximity to multiple brainstem nuclei and fibre tracts, progressive neurological decline is observed in 39% patients with infratentorial CMs.[2] Magnetic resonance imaging is the modality of choice, and susceptibility weighted imaging is the most sensitive means to detect blood products thus key to diagnosing cerebral CMs. Evolving blood products inside a CM appear as variable image intensities and give rise to the typical 'popcorn' appearance.[3] Since MRI appearance is usually pathognomonic, biopsy is rarely needed. Angiography is indicated only if MRI cannot exclude arteriovenous malformation. Genetic testing should be arranged to screen for familial cerebral CM. Treatment approach depends on the site, size, symptoms, and history of haemorrhage.[4] Indications for surgical resection of a cerebral CM include intracranial haemorrhage and epilepsy. Options include conventional surgery or stereotactic radiosurgery.[4,5]

Author contributions

All authors contributed to the concept or design, acquisition of data, analysis or interpretation of data, drafting of the manuscript, and critical revision of the manuscript for important intellectual content.

All authors had full access to the data, contributed to the study, approved the final version for publication, and take responsibility for its accuracy and integrity.

Conflicts of interest

As an editor of the journal, KL Hon was not involved in the peer review process. Other authors have disclosed no conflicts of interest.

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Ethics approval

Ethics approval for this study was obtained from Hong Kong Children's Hospital Ethics Committee, Hong Kong (Ref No.: HKCH REC 2019 009). The patient was treated in accordance with the tenets of the Declaration of Helsinki. Written consent for publication has been obtained from the patient's parent.

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