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Hypoplastic extraocular muscles due to sclerosing cavernous hemangioma of the cavernous sinus mimicking congenital fibrosis of the extraocular muscles

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Abstract

A girl with a clinical presentation consistent with unilateral congenital fibrosis of the extraocular muscles type 3 at 2 years of age years later developed progressive ophthalmoplegia and an afferent pupillary defect. Reimaging revealed a lesion diagnosed as a sclerosing cavernous sinus hemangioma with focal calcification. Cavernous sinus hemangioma is a rare, benign vascular malformation whose growth causes cranial nerve compression. Although frequently missed on routine neuroimaging, characteristic radiologic findings distinguish it from lesions such as meningioma.

Case Report

A healthy, 2-year-old girl presented Stein Eye Institute, University of California Los Angeles, with left mydriasis 8 days after striking her chin on a table. Born to nonconsanguineous parents, her birth and gestational history were unremarkable except for mild left ptosis since birth, which was also observed in her 8-month-old sister. On examination, she was normocephalic and had normal vision bilaterally. There was a 2 mm left blepharoptosis with eyelid crease present. Levator function was 12 mm bilaterally. She was orthotropic, but her left eye exhibited moderate limitation to supraduction, mild limitation to abduction and adduction, and absent Bell's phenomenon. The left pupil diameter measured 5 mm but was nonreactive to light. Slit-lamp and fundus examinations were normal bilaterally. There was no other neurologic abnormality.

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Noncontrast magnetic resonance imaging (MRI) of the brain and orbits performed without contrast (1.5-Tesla Signa; General Electric, Milwaukee, WI) revealed extraocular muscles of subnormal size in the left orbit (Figure 1A), and left hypoplastic intraorbital motor nerves. The diameter of the subarachnoid oculomotor nerve was 1.4 mm on the left but 2.0 mm on the right. The brain appeared normal. Forced duction testing under anesthesia demonstrated free elevation of both eyes. The initial impression was early congenital fibrosis of the extraocular muscles type 3 (CFEOM-3), without development of restriction. The family declined genetic testing. Congenital oculomotor nerve palsy was also considered.

Symptoms and signs were stable for the next 2 years. When the patient was reexamined at age 5 years, the parents reported adoption of a left head turn. Visual acuity measured 20/20 in the right eye and 20/80 in the left eye. There was a severe deficit of supraduction and a reduction in levator function to 7 mm in left eye (Figure 2). The patient was orthotropic at distance, with 4 of exophoria at near. There was a left afferent pupillary defect. Fundus and neurological examinations were unremarkable.

Brain and surface coil orbital MRI were repeated with and without contrast using published methods¹⁻³ and revealed progressive atrophy of the left subarachnoid oculomotor nerve to 1 mm diameter and further thinning of the left rectus muscles (Figure 1B). Within the left cavernous sinus, there was a heterogeneously enhancing mass measuring 12 mm anteroposteriorly by 5 mm transversely by 10 mm vertically (Figure 3A), containing numerous nonenhancing calcified nodules and consistent with phleboliths on X-ray computed tomography (Figure 3B). With this knowledge, a review of the initial MRI disclosed a similar-sized, indistinct tumor. The coarse calcifications extending into the left orbit were recognized to have progressed. There was no hypervascularity or tumor blush on cerebral angiography. The overall radiographic findings suggested sclerosing cavernous hemangioma of the cavernous sinus. The family declined neurosurgery.

Discussion

Our patient presented with blepharoptosis and ophthalmoplegia in the setting of familial ptosis. In light of hypoplasia of the oculomotor nerve, extraocular muscles, and intraorbital motor nerves, the initial diagnosis of CFEOM-3 was reasonable, assuming that the anticipated restriction due to contracture of normally innervated extraocular muscles had not yet emerged. CFEOM is characterized by nonprogressive ophthalmoplegia and ptosis. The atypical form CFEOM-3 is autosomal dominant, with incomplete penetrance and variable expression, and may be unilateral. It may result from missense mutations in *TUBB3*, encoding neuron neuron-specific β -tubulin.¹

MRI findings in CFEOM include variable, asymmetrical levator and superior rectus atrophy correlating with blepharoptosis, deficient supraduction, and small orbital motor nerves. Other extraocular muscles may exhibit variable hypoplasia correlating with duction deficit. Oculomotor nerve hypoplasia is typical.^{2,3} The nonreactive mid-dilated pupil and absence of restriction on forced duction testing in our patient were atypical of CFEOM-3. Congenital oculomotor nerve palsy has similar MRI findings of oculomotor nerve and extraocular muscle hypoplasia to those observed in our patient.¹⁻³ The diagnostic distinction between

this and CFEOM was initially regarded as unimportant because neither condition is directly treatable. However, clinical presentation of both CFEOM and congenital oculomotor nerve palsy are nonprogressive. Additional investigations were undertaken when progression was evident.

Cavernous hemangioma is a benign vascular malformation composed of hamartomatous blood vessels with thin-walled sinusoids without intervening neural tissue. The lesion may occur in any organ, including the central nervous system. Intracranial cavernous hemangioma occurs in 0.5% to 1% of the population, mainly in brain parenchyma. Only 2% are extra-axial, and of these, less than 2% involve the cavernous sinus.⁴⁻⁶

Typically occurring in females between the third and sixth decades of life, cavernous sinus cavernous hemangioma may impinge on nerves, resulting in neuropathy.⁵⁻⁷ Progressive blepharoptosis, ophthalmoplegia, diplopia, anisocoria, pain, and trigeminal hypesthesia have been reported.⁴⁻⁸ Visual acuity is reduced when extracavernous extension compresses the optic nerve.^{5,7} The youngest previously reported case of cavernous hemangioma was an 11-year-old boy.⁹

Specific diagnosis of cavernous sinus cavernous hemangioma is important because neurosurgery may induce uncontrollable hemorrhage, with mortality approaching 20%^{4,5,7} and iatrogenic cranial nerve palsies.^{5,6,10} MRI can distinguish cavernous hemangioma from more common tumors, such as meningioma. Typical features of cavernous hemangioma include prominent homogeneous hyperintensity with well-defined borders on T2-weighted MRI, contrast enhancement, and angiographic tumor blush.^{6,7,10} Tumor calcification in our patient distinguished it from typical cavernous sinus cavernous hemangioma, which normally lacks calcification. Our patient manifests the rare, sclerosing cavernous hemangioma that has, to our knowledge, been reported only once, in a 67-year-old woman with trigeminal neuralgia but no ophthalmoplegia.¹⁰

Symptom evolution in our patient followed the usual course of cavernous sinus cavernous hemangioma, typically insidious progression over weeks to 10 years.^{4,6} Left oculomotor nerve hypoplasia in our patient suggests that cavernous hemangioma was present congenitally. Hypoplasia of left extraocular muscles may have been due to tumor compression of orbital motor nerves. Decreased left eye visual acuity may be attributed to optic nerve involvement.

Treatment of cavernous sinus hemangioma depends on tumor size, extension, and vascularity. Modalities include preoperative embolization, electrothrombosis, piecemeal or en bloc resection, and radiosurgery.^{5,6,10}

Literature Search

PubMed was searched in the English language only in January 2014 for all articles published previously using the following search terms: *cavernous hemangioma of the cavernous sinus, cavernous hemangioma brain, cavernous sinus hemangioma, sclerosing cavernous hemangioma, extracerebral cavernous hemangioma, intracranial cavernous hemangioma, and extraaxial cavernous hemangioma.*

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FIG 1. Quasicoronal magnetic resonance imaging of the patient showing hypoplastic extraocular muscles in the left orbit at 2 years of age (A) and further progression of muscle atrophy at 5 years (B).

FIG 2. Clinical photographs of the patient at age 2 years (A), with progression at age 5 years (B), showing moderate to severe limitation to supraduction, and mild abduction and adduction deficits in the left eye.

FIG 3. Axial magnetic resonance imaging (A) and computed X-ray tomography (B) showing sclerosing left cavernous sinus hemangioma containing multiple coarse calcifications (arrows).