Case report

Small lymphaticovenous malformation of the orbital apex clinicopathologic correlation

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A B S T R A C T

Purpose: To familiarize clinicians with the clinical and magnetic resonance imaging (MRI) features of a small orbital apex lymphaticovenous malformation that resulted in blindness and evaded timely clinical diagnosis.

Observations: A 68-year-old man presented with severe vision loss due to a 9 mm mass at the apex of the orbit above the optic nerve. When surgically removed 4 years later, the lesion was characterized by vascular spaces of varying size. Larger ones were filled with fibrin and organized thrombi. Stromal septa of endothelial-lined cavernous spaces were partially necrotic and there was evidence of remote hemorrhage. Some endothelial cells expressed D2-40, a marker of lymphatic channels.

Conclusions and importance: Unless a high index of suspicion is maintained for a lymphaticovenous malformation the clinical diagnosis of a small but vision-threatening lesion can be overlooked.

1. Introduction

A subset of low flow vascular malformations of the orbit is characterized by an assemblage of endothelial cell-lined spaces of varying size that express vascular and lymphatic markers.\textsuperscript{1,2} Vascular malformations of this type are considered lymphaticovenous hamartomas, and may not present clinically until late in life. We report the clinical features of a small orbital apex lymphaticovenous malformation whose diagnosis remained obscure until it was removed surgically, four years after presentation. Lack of familiarity with the range of neuroradiologic findings associated with lymphaticovenous malformation may have contributed to this delay.

2. Case report

A 68-year-old man was referred for evaluation of poor vision in the left eye for three months. Vision loss was painless and occurred in the absence of any other associated symptoms. It could not be determined if vision loss occurred suddenly three months earlier or if it was unexpectedly noticed one day. Two magnetic resonance imaging (MRI) scans had been performed and both reported an abnormality at the left orbital apex; they were not available for review. The patient was otherwise healthy and past medical history was non-contributory.

Visual acuity on examination was 20/25 in the right eye and counting fingers at three feet in the left eye. A left relative afferent pupillary defect was present. Ocular motility was full and there was no proptosis. The remainder of the examination, including slit-lamp and dilated ophthalmoscopy, were unremarkable except for early cataracts. An MRI scan revealed a discrete cigar-shaped mass at the apex of the left orbit, immediately superior to the optic nerve. The tubiform lesion was 9 × 3.1 mm and compressed the optic nerve beneath it (Fig. 1A). On axial and coronal images, the mass was hyperintense to muscle and to normal optic nerve on T1-weighted images and did not enhance with gadolinium. The outer crescentic of the mass on coronal views, however, had modestly stronger signal intensity than its core (Fig. 1B). All routine laboratory studies were either normal or negative, including serum angiotensin converting enzyme. As the possibility of sarcoidosis could not be excluded, the patient was treated with 80 mg of oral prednisone for four weeks, without improvement in visual function.

The patient was monitored with serial MRI scans every four to six months for four years. There was no change in the pattern of MRI signals as the mass increased in diameter by 1 mm and length by 5 mm over four years. No new medical or systemic disorders developed over this period. Vision slowly declined until it was no light perception. At this point an excisional biopsy was performed.

The mass could not be removed intact. It and the optic nerve...
Fig. 1. (A) Axial magnetic resonance images (MRI) showing a cigar-shaped mass lying above and parallel to the optic nerve. (B) Coronal MRI reveals the circular mass (arrow) compressing the optic nerve (arrowhead). The superior rim of the mass displays greater signal intensity.

Fig. 2. Section of optic nerve cut roughly parallel to it longitudinal axis shows atrophy and chronic inflammation concentrating in pial septae (H & E, bar = 160 μm). Inset, shows chronic inflammation with hemosiderin-laden macrophages concentrating in and around a pial septa (H & E, bar = 200 μm).

Fig. 3. Sections from the mass above optic nerve. (A) The largest biopsy was a blood and fibrin filled cavity partially lined by endothelial cells. A fibrous capsule surrounds the mass. A compressed hemangioma is noted in the upper left quadrant (H & E, bar = 400 μm). (B) Another blood and fibrin-filled mass with surgically transected ends was festooned by partially necrotic and fibrin-covered septae (H & E, bar = 400 μm).}

beneath it were removed separately in several pieces. Histologically, the optic nerve was covered by pia mater. The axons were atrophic and the pial septa were chronically inflamed (Fig. 2). Hemosiderin-laden macrophages were plentiful (Fig. 2, inset). There were several large cystic spaces filled with blood, fibrin, and varying amounts of organizing thrombus (Fig. 3). The outer walls consisted of fibrous connective tissue (capsule) containing varying amounts of collagen. The largest cyst measured 3 mm in diameter (Fig. 3A). Another large blood-filled cystic cavity was festooned by hyalinized bands of collagen covered in fibrin (Fig. 3B). Many were partially lined by CD34-positive endothelial cells (Fig. 4). Thicker stromal septa were necrotic (Fig. 4B).

3. Discussion

We describe a relatively small lymphaticovenous malformation of the orbital apex that results in total vision loss. Lymphatic proliferations of the orbit are unique in the sense that they generally arise in tissue that normally lacks lymphatic channels. Two exceptions to this, however, are the lacrimal gland and optic nerve sheath.5,6 Their location in the optic nerve sheath can reasonably explain the potential origin of the hamartomatous proliferative seen in our patient. The pathogenesis of mixed lymphaticovenous malformations is not completely understood, but they appear to exist along a spectrum in which various proportions of venous and lymphatic channels coalesce.1–3,6–8 It is postulated that lymphatic endothelium in mixed lymphaticovenous malformations may develop from injured or dysmorphic vascular endothelium.9 Venous thrombosis could play a central role in triggering the events that lead to the emergence of lymphatic endothelium.

The diagnosis of lymphaticovenous malformation was not entertained in our patient clinically because the lesion did not demonstrate intermediate or hypointense signals on T1-weighted MRI for a lymphangioma.1 A pure cavernous venous malformation was not considered because the lesion was not isointense to muscle on T1-weighted images and did not enhance with contrast.10 Compared to the majority of lymphaticovenous malformations described in the literature, however, this lesion was small. Most of the volume was occupied by several spaces filled with fibrin and thrombi that pushed smaller caliber vascular and lymphatic spaces to the side. Given a lesion with maximum diameter of just over 1 cm and location at the apex of the orbit, the standard methods for hemodynamic assessment of lymphaticovenous malformation (e.g. Doppler ultrasound with and without Valsalva, Valsalva-augmented venous phase multidetector computer tomography angiography, etc.) may not have yielded insightful results.1,11

If confidently diagnosed, how might such a small vascularized lymphatic malformation be managed adjacent to the optic nerve without inflicting collateral injury? Space does not permit adequate review of this therapeutic challenge, but alternatives to surgical excision might include injection of sclerosants or ablation with carbon dioxide laser.1,12 Future pharmacological agents that inhibit lymphatic/venous endothelial cell proliferation might represent another option.1

In summary, we describe a lymphaticovenous malformation of the orbital apex whose natural history over four years resulted in vision loss. Perhaps because of its small size, MRI studies did not suggest the possibility of a low-flow vascular malformation. Optimal strategies for diagnosing and managing vascular lesions in this location are being developed.12 Awareness of the clinical spectrum of this condition should allow for earlier clinical recognition, a critical step in advancing therapeutic strategies.

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