PHOTO ESSAY

Bilateral Optic Nerve Head Angiomas and Retrobulbar Haemangioblastomas in von Hippel-Lindau Disease

Masoud Aghsaei Fard, Narges Hassanpoor, and Razieh Parsa

Farabi Eye Research Center, Department of Ophthalmology, Tehran University of Medical Sciences, Tehran, Iran

ABSTRACT

A 39-year-old man presented with progressive visual loss in both eyes. His past medical history was remarkable for biopsy-proven bilateral adrenal pheochromocytomas. Ophthalmoscopy revealed an optic nerve head angioma and macular lipid exudates in the right eye and a regressed optic nerve head angioma in the left eye. A fluorescein angiogram revealed a lobulated pattern of leakage and venous staining from the optic nerve head angioma. Brain and orbital magnetic resonance imaging showed haemangioblastomas of the right intraorbital and left paraclinoid optic nerves and in the left cerebellopontine angle area. These findings were consistent with a clinical diagnosis of von Hippel-Lindau disease.

Keywords: Haemangioblastoma, optic nerve, von hippel-lindau

CASE REPORT

A 39-year-old man presented with progressive visual loss from 2 years ago, first in his left eye and then, a year later, in his right eye. His past medical history was remarkable for biopsy-proven bilateral adrenal pheochromocytomas 20 years ago. He had been followed with a diagnosis of bilateral optic nerve glioma and left cerebellopontine angle schwannoma for 2 years based on his brain magnetic resonance imaging (MRI). In addition, he had twice received corticosteroid pulse therapy for possible demyelinating disorder over a 2-year period because of multiple periventricular plaques. His family history was remarkable for early death due to stroke in his sister. On neuro-ophthalmic evaluation, the best-corrected visual acuity was finger count OD and light perception OS. Pupils were equal in size but reacted sluggishly to light. There was a 1+ left relative afferent pupillary defect. Ophthalmoscopy revealed an optic nerve head angioma with macular lipid exudates in the right eye and a regressed optic nerve head angioma in the left eye. A fluorescein angiogram revealed early hyper-fluorescence and venous

staining from the right optic nerve head and late staining from the left optic nerve head (Figure 1). Brain and orbital MRI revealed three well-demarcated, intensely gadolinium-enhancing masses in the left cerebellopontine angle, the right intraorbital optic nerve, and the left paraclinoid optic nerve (Figure 2). Based on the imaging characteristics, a differential diagnosis of optic nerve glioma, meningioma, metastases, and granulomatous lesions was entertained and the top presumptive diagnosis was optic nerve gliomas. However, based on the bilateral optic nerve head angiomas and biopsy-proven diagnosis of pheochromocytoma, the MRI findings most likely were optic nerve and intracranial haemangioblastomas consistent with a diagnosis of von Hippel-Lindau (VHL) disease. In other words, the presence of multiple lesions (bilateral optic nerve head angiomas, bilateral optic nerve and posterior fossa haemangioblastomas, and pheochromocytomas) in this patient suggested a clinical diagnosis of VHL disease. Apart from an early stroke in his sister, there was no other family history suggestive of VHL and genetic testing was not performed in the family members. Patient refused to accept any treatment modality for his active

FIGURE 1 Right eye fundus photograph (A) shows stage 3 (exudative) optic nerve head capillary angioma with macular exudates. Angiography (B, C) reveal early hyper-fluorescence and venous staining in a lobulated pattern. Left eye fundus photograph (D) and angiography (E, F) show the stage 2 (classic), regressed optic nerve head capillary angioma with early hyper-fluorescence and late staining.

right optic nerve head angioma, such as photodynamic therapy or intravitreal bevacizumab. Chest and abdominopelvic computed tomographic (CT) scan was performed, which was normal. We explained the risk of renal cell carcinoma and recommended close monitoring.

COMMENTS

Von Hippel-Lindau is a neurocutaneous syndrome that is transmitted by autosomal-dominant pattern with incomplete penetrance. Because of mutation in the VHL tumour suppressor gene, this disease is associated with increased risk of various benign and malignant tumours, including multifocal central nervous system (CNS) and optic nerve

haemangioblastomas, retinal angioma, pheochromocytoma, and renal cell carcinoma.^{1,2} Most of the CNS haemangioblastomas present as posterior fossa masses. In this case, we observed cerebellopontine haemangioblastoma. Although retinal angiomas are frequently present in patients with VHL, capillary angioma of optic nerve head similar to our case is less common. On the other hand, optic nerve haemangioblastomas are extremely rare and about 20 cases of them have been reported in the literature so far. Most of them were associated with VHL disease and were located within the intraorbital and/or intracranial portions of the optic nerve.³⁻⁵ Kerr et al. described a case of VHL disease with progressive visual loss in the right eye. The patient had a small capillary angioma of the right optic disc and a vascular mass of the intracranial portion of the right optic nerve that

FIGURE 2 Brain magnetic resonance imaging shows right intraorbital and left cerebellopontine angle tumours on T1-weighted imaging (A, black arrows). T1-weighted imaging (B) shows gadolinium-enhancing lesion in the left paraclinoid optic nerve.

was found to be a haemangioblastoma. Her vision had changed from 20/400 to count fingers after complete resection by microdissection, and the tumour resection was recommended as the optimum treatment for haemangioblastoma.⁵ The location and shape of the intracanalicular optic nerve lesion in that study was similar to our patient's left optic nerve haemangioblastoma. Another case report presented a female who had previously lost vision secondary to a retinal haemangioblastoma. Ipsilateral optic nerve haemangioblastoma produced optic tract oedema, which resolved after resection of the prechiasmal tumour.⁶ The preoperative differential diagnosis for this tumour based on imaging studies has included glioma, meningioma, and neurilemoma ("neurilemoma"). Unlike gliomas, haemangioblastomas enhance brightly with gadolinium as in our case, allowing differentiation of these tumours.

Similar to haemangioblastomas, meningiomas enhance; however, the enhancement arises from the nerve sheath.⁵ To our knowledge, only one patient with bilateral optic nerve haemangioblastomas has been reported, by Ginzburg *et al.* in 1992. That bilateral tumour involved the intracanalicular prechiasmal portion of the optic nerves and caused blindness.⁷

Therefore, the presence of retinal or optic nerve head angioma should prompt clinicians to consider and investigate the possibility for VHL disease, which can be associated with life-threatening conditions such as renal cell or clear cell carcinoma and pheochromocytoma. Surveillance protocols for monitoring patients and family members at risk have been developed.^{8,9}

In conclusion, although haemangioblastomas of the optic nerve are rare, a high index of suspicion is warranted in any patient with signs of VHL, such as pheochromocytoma or cerebellar haemangioblastoma. Vision loss in VHL may also be due to retinal lesions, but neuro-ophthalmological evaluation may raise suspicion for optic nerve haemangioblastoma.⁶ As in our patient, contrast enhanced MRI of the orbit and the brain is helpful in these cases.

Declaration of interest: The authors report no conflicts of interest. The authors alone are responsible for the content and writing of the paper.

Note: Figure 1 of this article is available in colour online at <http://informahealthcare.com/oph>.

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