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CASE REPORT

Von Hippel-Landau disease presenting as a retinal hemangioblastoma: A case report

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Abstract

An 18-year-old female presented with outward deviation of the right eye (RE) for 8 years. She gave history of low vision in RE since childhood and vision were denies PL in RE on presentation. Vision in the left eye (LE) was 6/9. Patient had records of undergoing fundus fluorescein angiography and laser photocoagulation of a retinal angioma in LE in 2011 at a private hospital where she was also diagnosed with exudative retinal detachment with complicated cataract in RE. Fundus examination of LE showed a lasered angioma along the superotemporal arcade and a retinal angioma in the inferotemporal arcade. Laser barrage of the retinal angioma in LE was done and patient operated for cosmetic squint correction in RE. Magnetic resonance imaging brain showed a hemangioblastoma in the RE and one in the fourth ventricle of the brain. Ultrasound abdomen was normal.

Introduction

Retinal capillary hemangioblastomas, often referred to as retinal angiomas, is typically an indication of the Von Hippel-Lindau (VHL) illness. They may also appear as a stand-alone entity apart from systemic disease. It is the earliest and most typical symptom of VHL disease. Every case of retinal angiomas should undergo a complete fundus examination to find all angiomas and a complete systemic evaluation to look for other manifestations of the disease.[1]

Case Report

An 18-year-old female presented with outward deviation and low vision of the right eye (RE) for 8 years. Patient gave history of being diagnosed with retinal angioma in the left eye (LE) in 2011 for which fundus fluorescein angiography (FFA) was done which showed an area of leakage ½ DD outside the inferotemporal arcade following which a laser photocoagulation of the angioma was done. Exudative retinal detachment (RD) and complicated cataract was noted in RE in old records.

Vision in RE was PL negative and LE was 6/9.

On examination, RE showed 20° exotropia which did not take fixation on cover uncover test. Anterior segment showed total cataract with 360° posterior synechia with no view of fundus ultrasound (USG) examination of posterior segment revealed a funnel-shaped RD.

Anterior segment examination of LE was WNL. Posterior segment examination showed a lasered retinal angioma along the inferotemporal quadrant and a retinal angioma along the superotemporal arcade. FFA was done which showed area of leakage around the angioma. Laser barrage was done in multiple sitting.

A full body work up was also done to look for tumors in other parts of the body as part of the spectrum of VHL syndrome. Magnetic resonance imaging (MRI) brain showed an intensely enhancing lesion in caudal aspect of fourth ventricle measuring 5 × 6 mm with prominent adjacent vessels and another intensely enhancing lesion in posterior chamber of the right ocular globe of 9.8 × 10.8 mm with prominence of vessels along the posterior aspect of the lesion arising from the right lateral and inferior wall suggestive of hemangioblastoma. USG whole abdomen done

to look for renal cell carcinoma/cyst, pheochromocytoma, and pancreatic tumor/cyst was normal.

Patient then underwent squint surgery in RE for cosmetic squint correction.

The patient's two other siblings were screened for retinal hemangioblastomas and both did not show any evidence of the disease.

Discussion

The retinal hemangioblastoma is the earliest and most common manifestation of VHL disease. It is a vascular hamartoma and its clinical manifestations often begin within the first two decades of life. VHL syndrome is accompanied by bilateral or multiple retinal hemangioblastomas. Solitary retinal hemangioblastomas may also be associated with VHL syndrome.

The VHL gene, a tumor suppressor gene located on the short arm of chromosome 3 (3p25–26), is the cause of VHL due to a germ line mutation.

VHL gene is a tumor suppressor gene found on the short arm of chromosome 3 (3p25–26). A germline mutation in this gene causes VHL disease.[2]

Lesions can be peripheral or juxtapapillary. Diagnosis is mainly clinical. A thorough fundus examination is a must as multiple angiomas may be present.

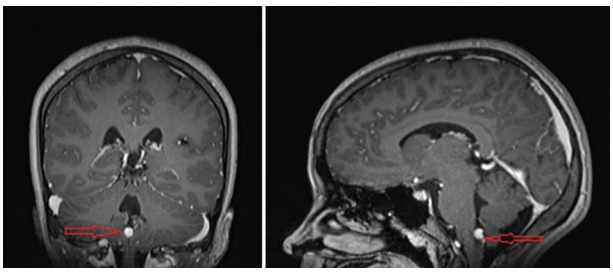
Hard exudates and retinal edema are frequently observed with the tumor and mostly involve the macula. Juxtapapillary lesions, which are present in about 11–15% of patients, can elevate and exude around the optic nerve, resulting in pseudopapilledema, and they can be the only sign of retinal VHL illness. In <3% of instances, retinal or vitreous hemorrhages are detected.

Hemangioblastoma is a vascular hamartoma. These lesions can be peripheral or juxtapapillary, seen in 11–15% cases. They are usually associated with retinal edema and hard exudates which often can involve the macula also. Juxtapapillary lesions may cause pseudo papilledema which may even be the only manifestation of retinal disease. They may rarely be associated with retinal or vitreous hemorrhages.[3]

The investigation of choice for a retinal hemangioblastoma is a FFA. There is a rapid filling of the feeding artery followed by filling of the tumor mass then a rapid exit from the draining vein. Pinpoint tumors that may not be detected clinically can be detected on FA.[4]

In eyes with significant RD, MRI and computed tomography (CT) can reveal an enhancing retinal mass. These scans are especially crucial for identifying associated abdomen and CNS tumors in VHL syndrome. Depending on whether an at-risk relative or a genetically positive patient is being evaluated, different techniques are employed to assess the systemic symptoms of VHL.

MRI and CT may show enhancing retinal masses in the eye with extensive RD. These scans can also be used to detect associated central nervous system and abdominal tumors in VHL syndrome. Depending on whether the genetically positive patient or at-risk relative is being examined, multiple protocols are used for the evaluation of VHL systemic features.[4]



Small, non-visually-threatening peripheral lesions that are <500 um in diameter and do not have an accompanying exudate can be observed. Nearly, all retinal hemangioblastomas need to be treated since VHL syndrome-related tumors have a tendency to be more aggressive. Small lesions (3 mm) can be treated with laser photocoagulation or photodynamic therapy (PDT); medium lesions (3–6 mm) can be treated with PDT or cryotherapy; and big lesions (>6 mm) can be treated with PDT, plaque irradiation, or internal resection through the pars plana vitrectomy method.[5-7]

Conclusion

Hemangioblastoma is a vascular hamartoma. Juxtapapillary lesions may cause pseudo papilledema which may even be the only manifestation of retinal disease.

FA is the best test for identifying and validating retinal hemangioblastoma because it demonstrates a quick filling of the feeding artery, followed by the tumor, and a rapid evacuation through the draining vein. Small pinpoint tumors that may not be detected clinically can be diagnosed on FA.[4]

Clinical Significance

The earliest and most common manifestation of VHL disease is a retinal hemangioblastoma. It usually manifests in the first two decades of life. Prompt diagnosis can be made with fundus evaluation.

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