

Von Hippel Angioma

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SUMMARY

We report a three year follow up of a 35 year old Indian gentleman who presented with sudden, painless blurring of left (L) eye vision with initial visual acuity (VA) of 6/60. Fundoscopy revealed (L) vitreous haemorrhage and subsequently confirmed a (L) inferotemporal capillary haemangioma. The adjacent area of capillary haemangioma was treated with barricade argon laser photocoagulation to prevent progression of exudative retinal detachment inferiorly. Subsequent follow up showed mild regression of capillary haemangioma with maintenance of (L) eye vision at 6/9.

KEY WORDS:

Retinal angioma, von Hippel angioma, von Hippel Lindau syndrome

INTRODUCTION

Retinal angioma is a benign potentially blinding vascular hamartoma, frequently associated with the dominantly inherited familial cancer syndrome, von Hippel Lindau (VHL) disease. It is the most frequent and earliest manifestation of VHL disease, usually diagnosed in 2nd to 3rd decade of life. Retinal angioma commonly occurs in the midperiphery of retina as an orange-red, spherical well circumscribed tumour supplied by enlarged and tortuous feeder vessels emerging from the disc and leading to the tumour¹. Although it is a rare disease with potential systemic involvement, it can be devastating both to the affected individuals and possibly to the rest of their family, necessitating regular surveillance to detect subtle signs of life threatening complications, such as central nervous system haemangioblastoma, pheochromocytoma or renal cell carcinoma².

CASE REPORT

A 35 year old Indian man presented with sudden, painless blurring of left (L) eye vision without history of trauma or family history of ocular disease. Initial visual acuity (VA) was 6/60 OS, 6/6 OD. Fundoscopy showed frank (L) vitreous haemorrhage. Ultrasonography of left eye revealed vitreal hyperechoic areas suggestive of vitreous hemorrhage without evidence of retinal detachment. Anterior segment examination including intraocular pressure measurement of (L) eye was normal. Systemic reviews were unremarkable. He was treated conservatively and his left VA improved to 6/12 upon resolution of vitreous haemorrhage. As the vitreous hemorrhage resolved, the fundus examination showed (L) midperipheral inferotemporal retinal angioma measuring 1.5

disc diameters (DD) with 3 dilated, tortuous feeder vessels from the optic disc. There were fibrotic membranes around the dilated feeder vessels and the retinal angioma with adjacent posterior vitreous detachment (Figure 1).

Hence, a provisional diagnosis was von Hippel angioma¹, with probable underlying von Hippel-Lindau disease. Fundus fluorescein angiography and cryotherapy were not performed as the patient did not consent for any invasive procedure as his vision improved. He consented to barricade the lesion with argon laser photocoagulation in the hope of preventing formation of adjacent subretinal exudates. Despite laser photocoagulation, he developed a localized shallow exudative retinal detachment inferiorly but remained stable. Ultrasonography showed acoustically solid retinal mass of medium reflectivity with a smooth anterior border and surrounding shallow retinal detachment (Figure 2).

On follow-up, his vision remained stable at 6/9. Although at present the workup (genetic studies pending) did not show positive evidence for von Hippel-Lindau disease, he has been put under regular follow up for three yearly MRI brain, spinal cord and yearly ultrasound abdomen, 24H urine catecholamine surveillance.

DISCUSSION

Retinal angioma consists of capillaries with fenestrations which secondarily lead to subretinal exudation. Rarely does it present as retinal or vitreous haemorrhage which is in contrary to this case. It may occur sporadically in the retina, termed von Hippel angioma but more often appears in VHL disease up to 70%¹. Regardless of its feared systemic association such as central nervous system haemangioblastoma, renal cell carcinoma, pheochromocytoma, adenoma, angiomas and cysts of kidney, liver, pancreas and epididymis, the retinal angioma appears identical clinically and histopathologically.

Patients presenting with a solitary retinal angioma as a single manifestation represents a clinical dilemma. Retinal angiomatosis is the first apparent lesion in most patients; underlying VHL disease may not be evident at diagnosis of ocular disease. With recent advances in molecular genetic screening, defect in VHL tumour suppressor gene at human chromosome 3 at locus 3p25-26 could be identified to assist in diagnosis of VHL disease³. It is invaluable in patients with solitary VHL like angioma, an absence of family history and no evidence of systemic disease. However, ocular lesions may be sporadic and not associated with genetic mutations.

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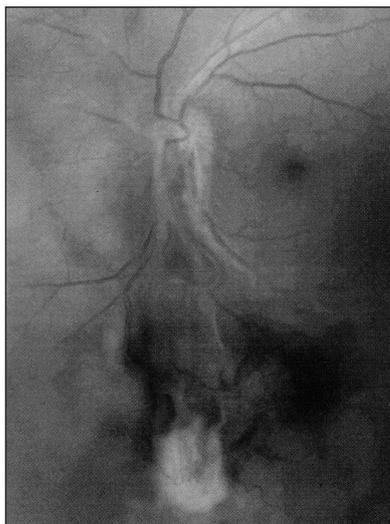


Fig. 1: von Hippel angioma with surrounding exudative retinal detachment and dilated feeder vessels extending from optic disc

Webster and coworkers did not find a mutation of VHL gene in 17 patients with solitary angioma in which they attributed them to the presence of de novo mutation, incomplete penetrance or inadequacy of current available technique of DNA analysis³. Screening for gene mutations in at risk individuals allows the identification of carriers among family members who may not yet manifest VHL disease in which they will be entered into a surveillance protocol. Interesting enough, there are investigators who hypothesized that electrophysiological dysfunction of inner retinal layer is present in individuals with VHL disease even without retinal angioma. This patient was subjected for lifelong comprehensive surveillance for systemic complications as it represents the safest option until genetic knowledge of all possible mutations is available.

Various methods for ablating retinal angioma have been previously reported: mainstays of treatment are laser photocoagulation and cryotherapy depending on tumour size and location¹. Brachyradiotherapy has also been advocated for lesions larger than 3.5mm to 4mm. Recent studies have also reported variable results with transpupillary thermotherapy, photodynamic therapy and vascular endothelial growth factor (VEGF) inhibitor SU5416¹. But in

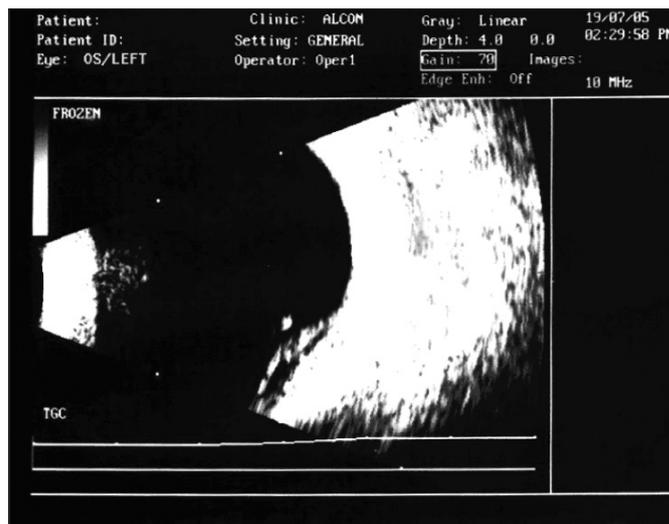


Fig. 2: B Scan showing shallow inferior retinal detachment surrounding hyperechoic retinal mass

this patient, the angioma was stable even without any treatment directly applied to a lesion of that size.

Prognosis for vision depends on tumour location, size and exudation but generally is excellent for treatment of tumour up to 1 disc diameter in size. Larger tumours are more difficult to treat and have a more uncertain treatment outcome.

CONCLUSION

Von Hippel angioma is a rare but potentially fatal disease with systemic involvement. Therefore, physicians and ophthalmologists should be familiar with this condition and manage the affected individuals and possibly to the rest of their family with high index of suspicion to detect subtle signs of life threatening complications.

REFERENCES

1. Gauthier D, D'Amico DJ, Mukai S: von Hippel -Lindau Disease. *Int Ophthalmol Clin* 2001; 41(4): 173-87.
2. Maher ER, Yater JR, Harries R *et al*. Clinical features and natural history of von Hippel-Lindau disease. *Q J Med* 1990; 77: 1151-163.
3. Webster AR, Maher ER, Bird AC *et al*. A Clinical and Molecular Genetic Analysis of Solitary Ocular Angioma. *Ophthalmology* 1999; 106: 623-29.