

BILATERAL OPTIC NERVE HEAD HEMANGIOMA IN VON HIPPEL LINDAU DISEASE: REPORT OF A CASE WITH SEVERE VISUAL LOSS

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Abstract - A 22 year-old woman presented with gradual visual loss in her right eye since 1990. Medical and family histories were unremarkable. Her visual acuity was 20/80 and 20/20 in right and left eyes respectively. Slit lamp examination was quite normal with no relative afferent pupillary defect. Fundi of both eyes revealed subretinal lesion with optic nerve head involvement and subretinal fluid in papillomacular bundle with macular pucker in right eye. Whole body MRI revealed a large hemangioma in the spinal canal. In 1998 the patient had significant visual loss at both eyes. This is the first report of Von Hippel disease with bilateral optic nerve hemangioma in Iran. *Acta Medica Iranica* 39 (2): 126-129; 2001

Key Words: Von-Hippel Lindau, optic nerve hemangioma

INTRODUCTION

Von Hippel Lindau (VHL) is one of the phacomatosis (1), the gene of which lies over the distal part of the short arm of chromosome 3 (2). Patients usually present with ocular complications in the second or early third decade; tumors may be found anywhere in globe from the disc to ora serrata. Involvement of the optic disc by itself occurs very rarely (3). Whenever there is only hemangioma on the retina or optic disc, the entity is called Von Hippel disease, but if there is CNS or visceral hemangioma or other malformation, it is not called VHL (4).

CASE SUMMARY

A 22 year old woman was referred to my office, because of gradual visual loss of her right eye in 1990. Routine eye examinations were done. Visual acuity was 20/80 and 20/20 in her right and left eye respectively with no relative afferent pupillary defect. In funduscopy exam a subretinal mass was found around the optic nerve head of both eyes and subretinal fluid in papillomacular bundle with macular pucker in the right eye.

Angiography of both eyes was done (Fig. 1). MRI revealed hemangioma in spinal cord, at the level of L₁ to L₄ (Fig. 2). Laser spots were applied at the

boundaries of the right eye lesion; treatment within 100 to 200 μ m of the optic nerve was not performed to lower the chance of thermal necrosis of disc tissue, and at least 1.5 clock hours of the papillomacular bundle on the temporal side of the disc was spared of photocoagulation. And in the foveal side the laser spots were within 500 μ m of foveal center. No treatment was done for her left eye. Despite this treatment, laser spots were not effective in prevention of exudative retinal detachment and subretinal fluid accumulated (Fig. 3) and after several years, the visual acuity of her right eye became NLP LP respectively, and fundus examination, revealed bilateral exudative retinal detachment with neovascular glaucoma in right eye.

DISCUSSION

Optic nerve hemangioblastoma should be considered in the differential diagnosis of optic nerve tumors in patients with or without VHL disease (5). Angiomas affecting the optic nerve head and peripapillary retina maybe more difficult to recognise because of absence of visible afferent and efferent dilated vessels (6). As in this case, twenty percent of these patients will develop central nervous system tumors (hemangioblastomas of the cerebellum, medulla, pons, and spinal cord) (6). MRI screening should be considered for all patients with VHL gene to detect the multiple lesions in this disease (7). In MRI a large hemangioma in the spinal cord was detected in our patient. Optic disc tumors maybe asymptomatic. Visual loss results from intraretinal exudation, macular edema or surface wrinkling retinopathy (8).

Treatment is hazardous and may be associated with a temporary but marked increase in the amount of subretinal fluid and occasionally may result in the development of a total retinal detachment (6). When an ocular lesion is discovered, a systemic workup should also be considered for possible renal cell carcinoma and pheochromocytoma as well as for central nervous system lesions (6).

In conclusion, although optic nerve head hemangioma is a rare disease, it's a devastating disorder with no definite treatment. This is the first report of bilateral optic nerve head hemangioma in VHL.



Fig. 1a. Hemangioma of right optic nerve head in red-free picture

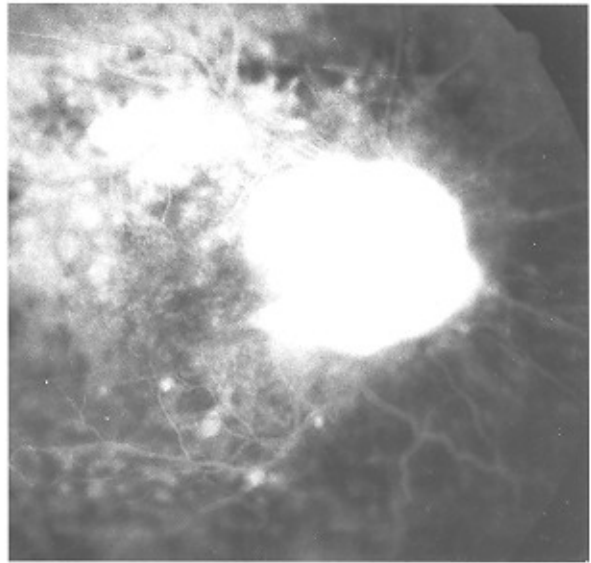


Fig. 1c. Fluorescein angiography of the right eye with dye leakage from hemangioma



Fig. 1b. Hemangioma of left optic nerve head in red free picture

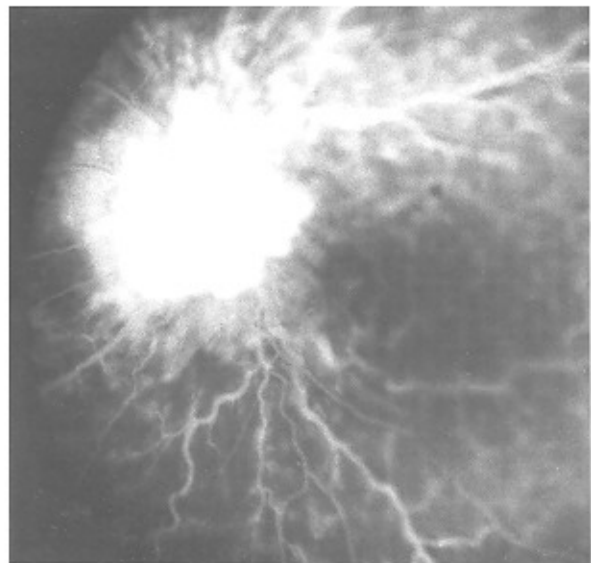


Fig. 1d. Fluorescein angiography of left eye with dye leakage from left optic nerve head



Fig. 2. MRI of spine Hemangioma at the level of T₄

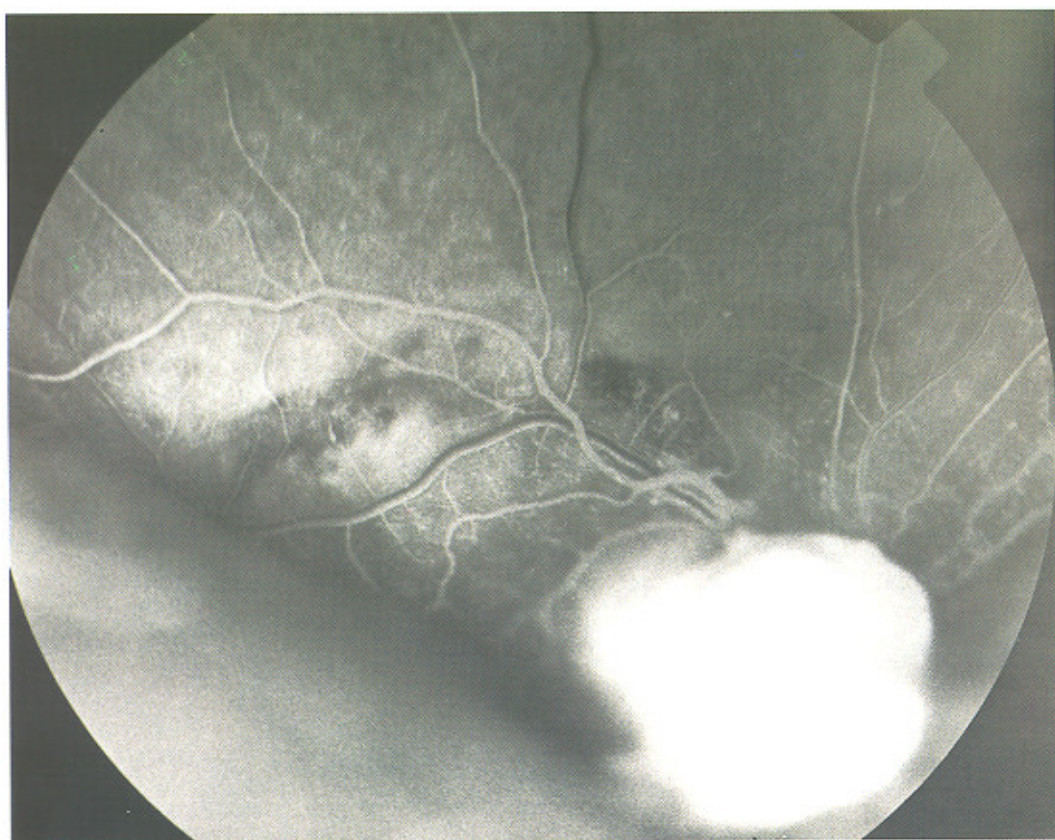


Fig. 3. Fluorescein angiography of the right eye with dye leakage from hemangioma and inferior retinal detachment.

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