AN OPTIC NERVE TUMOR IN VON HIPPEL-LINDAU DISEASE, MASQUERADING AS A RETINAL HEMANGIOMA

ANGIOMA RETINIANO COMO SÍNDROME MASCARADA DE TUMOR DEL NERVIO ÓPTICO EN LA ENFERMEDAD DE VON HIPPEL-LINDAU

FONS MARTÍNEZ MR¹, ESPAÑA GREGORI E², AVIÑÓ MARTÍNEZ JA², HERNÁNDEZ PARDINES F¹

ABSTRACT

Clinical case: A 35-year-old man with a family history of von Hippel-Lindau disease was diagnosed to have two retinal hemangiomas in the right eye and another in the left eye. The hemangiomas were treated with cryotherapy and laser photocoagulation respectively. Despite apparent good resolution of the retinal lesions, progressive visual loss was observed. An MRI was then performed and showed bilateral tumoral lesions of the optic nerve compatible with a glioma, meningioma or hemangioblastoma. Currently the visual acuity in his right eye is hand movement, and is 0.6 in the left eye.

Discussion: The presence of the retinal hemangiomas delayed the diagnosis of an optic nerve tumor in this patient (Arch Soc Esp Oftalmol 2006; 81: 293-296).

Key words: Von Hippel-Lindau disease, retinal hemangioma, optic nerve tumor, optical hemangioblastoma.

Received: 1/4/05. Accepted: 18/5/06.
¹ Ph.D. in Medicine.
² Graduate in Medicine.
Communication partially presented at the LXXX Congress of S.E.O. (Córdoba 2004).

Correspondence: M.ª Rosario Fons Martínez
C/. Maestro Sosa, n.º 18, pta. 30
46007 Valencia
Spain
E-mail: rosfomar@hotmail.com

SHORT COMMUNICATION

RESUMEN

Caso clínico: Varón de 35 años con antecedentes familiares de enfermedad de von Hippel-Lindau (VHL) diagnosticado de dos hemangiomas retinianos en ojo derecho (OD) y uno en ojo izquierdo. Las lesiones se trataron con crioterapia en OD y láserterapia en OI. A pesar de una aparente buena evolución de las lesiones retinianas hubo una progresiva disminución de la AV. Se realizó RM donde se evidenció lesiones tumorales del nervio óptico (NO) bilaterales compatibles con glioma, meningioma o hemangioblastoma. Actualmente: AV (OD): movimiento de manos y AV(OI): 0,6.

Discusión: La presencia de los hemangiomas retinianos en este paciente hizo retrasar el diagnóstico de un tumor en el NO, infrecuente en esta entidad.

Palabras clave: Enfermedad de von Hippel-Lindau, hemangioma retiniano, tumor del nervio óptico, hemangioblastoma del nervio óptico.
INTRODUCTION

Von Hippel-Lindau’s disease (VHL), also known as retinal-cerebellum angiomatosis, is a multi-systemic disorder with a preference for the retina and the central nervous system (CNS). The estimated prevalence of this disease is of 1 in 36,000 births (1).

The cause of this disease lies in a mutation of the VHL tumor suppression gene located in chromosome 3p25-26, with dominant autosomic inheritance with irregular penetration. It is characterized by a high degree of intra- and inter-family clinical variability (2).

The extraocular expressions of VHL (3) include CNS hemangioma, kidney cysts and adenocarcinomas, pheochromocytoma, cysts and tumors of the insular cells of the pancreas, epidimiry cystadenomas and tumors of the endolymphatic sac. Likely ophthalmological expressions are (3) capillary retinal hemangioma, other retinal vascular hamartomas and the retinal «twin vessels» (coupled venule and arteriole, separated by less than a venule diameter), whereas the involvement of the optic pathway due to a CNS is extremely rare.

In a VHL case report we present the joint prevalence of two ophthalmological findings: retinal hemangioma, which is very frequent in this entity, and the optic nerve tumor which is extremely infrequent.

CASE REPORT

A 35-year old male whose father and brother are affected by VHL, who came for an eye fundus exploration complaining of a slightly blurred vision in the right eye and exhibited no additional associated ophthalmological or systemic symptoms at the time.

The initial exploration gave a visual acuity (VA) of 1 in both eyes, with presence of two retinal hemangiomas in the right eye (RE) with slight exudation. The rest of the ophthalmological exploration gave normal results. The systemic study revealed a kidney cyst, without other findings. The cranial CAT was normal.

Six months later, the patient returned due to VA reduction in the RE (VA=0.6). a Fluorescein angiography was performed which evidenced a macula epi-retinal membrane with small exudation associated to the retinal hemangiomas (fig. 1). A brain MR was made with normal results. The patient was treated with transconjunctival cryotherapy for the retinal lesions.

One year later, VA in the RE was of finger-counting at 50 cm regardless of an apparently positive response to the treatment.

Two years later the patient referred loss of eyesight in the LE which evidenced a peripheral retinal angioma without exudation, treated with photocoagulation. A brain MR was performed, evidencing lesions in both optic nerves with intense hyper-capituring of contrast (more in the right optic nerve, where it reached the chiasmatic area). Said lesions were radiologically compatible with meningiomas, gliomas or even hemangioblastomas (figs. 2-4).

Four years later, VA in the RE was hand movement and 1 in the LE. At that time, the patient was diagnosed with a pheochromocytoma. Regular eye fundus checks and orbitary and cranial MR continue to be performed. The neurosurgery service discarded the possibility of an operation to preserve the remaining eyesight, which excludes the histopathological confirmation of the lesions.

At this time, seven years after beginning the process, the RE is amaurotic and the eye fundus exhibits scars of the old retinal hemangiomas, retinal folds and papillary paleness. In the LE, VA is of 0.6 and the treated hemangioma can be seen in the temporal area, as well as papillary paleness. The visual
field of the LE exhibits a central defect which extends to the nasal hemi-field (fig. 5).

DISCUSSION

Retinal capillary hemangioma is the most frequent ophthalmological expression (frequently the first) of Von Hippel-Lindau’s disease.
It is usually located in the superior temporal (39%) or inferior temporal (27%) peripheral retina. Juxta-papillary hemangioma has been referred in 11-15% of VHL cases (3).

According to previous studies, von Hippel-Lindau’s disease associates hemangiomas in the cerebellum in 35-59% of patients, and in the spinal medulla in 13-14%. Additional localizations of the central nervous system are infrequent and rare in the retrobulbar portion of the optic nerve (3,4).

In 1988, Nerad et al (5) published a case of optic nerve hemangioblastoma and carried out a bibliographic review, collecting six previous cases of retrobulbar optic nerve hemangioma. Of the seven cases, four are associated to von Hippel-Lindau’s disease. The present case is a patient with VHL’s disease exhibiting two tumors depending from the retrobulbar portion of both optic nerves. The X-ray characteristics are more reminiscent of meningioma or glioma. However, in the absence of an anatomic and pathological exploration, hemangioblastoma cannot be discarded. Said exploration is not available in this case because the Neurosurgery Service decided to adopt a «wait and see» approach, excluding surgery for the time being. In fact, the literature describes cases (4,5) in which tumors with a first X-ray based diagnostic of meningioma were subsequently diagnosed as heman-gioblastoma after the anatomic-pathological study. Both gliomas and meningiomas are solid tumors with intense contrast hyper absorption. Hemangioblastoma is usually a cystic tumor with a hyper-absorbing nodule, but it can also be solid with intense hyper-absorption and, when this occurs, it is difficult to differentiate from the previously mentioned tumors in X-rays.

The diagnosis of hemangioblastoma is supported by the fact that this patient has a prior diagnostic of VHL, a disease associated to the presence of hemangioblastomas in the CNS, and the fact of being male because there hemangiomas exhibit a preference for males, while meningiomas prefer females (4).

In this case it is also worthy pointing out the complex patient management. An extremely careful ophthalmological exploration must be performed, because the possibility of other coexisting lesions cannot be ignored. In this case, the existence of retinal angiomas, frequent in this entity (in addition to an initially normal MR) led to delays in diagnosing the main cause of the patient’s eyesight reduction, i.e., tumors in the retrobulbar portion of the optic nerve, a very exceptional occurrence in these cases.

**REFERENCES**