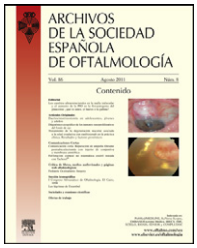
ARCHIVOS DE LA SOCIEDAD ESPAÑOLA DE OFTALMOLOGÍA

www.elsevier.es/oftalmologia



Short communication

# Retinal capillary hemangioma and von Hippel-Lindau disease: Diagnostic and therapeutic implications☆,☆☆

*R. Salazar*, C. González-Castaño, P. Rozas, J. Castro*

*Servicio de Oftalmología, Hospital Universitario Central de Asturias, Oviedo, Principado de Asturias, Spain*

ARTICLE INFO

*Article history:*
Received 2 February 2011
Accepted 8 March 2011
Available online 19 December 2011

*Keywords:*
Retinal capillary haemangioma
Von Hippel-Lindau disease
Laser photocoagulation
Vitrectomy

ABSTRACT

*Clinical case:* Man carrier of the von Hippel-Lindau (VHL) gene, with long-onset loss of vision in left eye. He had a retinal capillary haemangioma (HCR) and diffuses cystic edema in posterior pole. The systemic study revealed bilateral kidney tumors. Laser photocoagulation was performed which produced a subretinal and vitreous hemorrhage that required vitrectomy.
*Discussion:* Retinal capillary haemangioma (HCR) is the earliest and most frequent manifestation of the von Hippel-Lindau disease. Its detection requires it to be treated early and to rule out other visceral lesions. Laser photocoagulation is the most recommended treatment of small-size HCR. The most frequent complications are vitreous and subretinal hemorrhages.



## Hemangioma capilar retiniano y enfermedad de von Hippel-Lindau: implicaciones diagnósticas y terapéuticas

*Palabras clave:*
Hemangioma capilar retiniano
Enfermedad von Hippel-Lindau
Fotocoagulación láser
Vitrectomía

RESUMEN

*Caso clínico:* Varón con disminución de visión en ojo izquierdo de larga evolución. Portador del gen de VHL. Presenta hemangioma capilar retiniano (HCR) y edema quístico difuso en polo posterior. El estudio sistémico revela la existencia de masas renales bilaterales. Se inicia fotocoagulación con láser argón, produciéndose hemorragia subretiniana y hemorragia vítrea que precisó vitrectomía.
*Discusión:* El HCR constituye la manifestación más frecuente y precoz en la enfermedad de VHL. Su detección obliga a su tratamiento precoz, así como a descartar otras lesiones viscerales. La fotocoagulación láser es el tratamiento de elección en el HCR de pequeño tamaño. Entre sus complicaciones destaca la hemorragia vítrea y subretiniana.



☆ Partially presented as a clinical case panel at the 86 Congreso Nacional of the Sociedad Española de Oftalmología, September 22–25, 2010, Madrid, Spain.
☆☆ Please cite this article as: Salazar R, et al. Hemangioma capilar retiniano y enfermedad de von Hippel-Lindau: implicaciones diagnósticas y terapéuticas. Arch Soc Esp Oftalmol. 2011;86:218–21.

* *Corresponding author.*
E-mail address: raquelsalazarmendez@gmail.com (R. Salazar).

2173-5794/$ – see front matter 

## Introduction

The von Hippel-Lindau (VHL) disease is a hereditary disease caused by germinal mutations in the VHL tumor suppressing gene. The prevalence is estimated at 1/36,000 births and penetration is nearly complete by age 65. Approximately half of VHL cases are familial, with the other half being sporadic due to new mutations.[1,2]

The clinical expressions of VHL are highly diverse, with more than 40 lesions being described in 14 different organs, including retinal capillary hemangioma (RCH), cerebellum or spinal hemangioblastoma, renal pheochromocytomas and carcinomas (the main cause of death). Even though 50% of patients exhibited only one characteristic and very few develop the complete syndrome, all must undergo a systematic detection protocol (Table 1).[1,3]

Frequently, RCH is the first expression and is present in 70% of patients, with its frequency increasing with age. RCH can appear in isolation or as a part of the VHL disease, where one-third are multiple and half are bilateral. In the presence of familial history, the existence of RCH is considered to be a diagnostic criteria for the syndrome (Table 2). Possible ophthalmological expressions also include other vascular hamartoma and what is known as "twin vessels". Optic pathway alterations are rare, although one case has been described with an optic nerve tumor.[1–4]

**Table 1 – Von Hippel-Lindau disease detection protocols.**

| Assessment | Hawaii | Newfoundland | Cambridge |
|---|---|---|---|
| Ophthalmoscopy | Every 1–5 years from age 6 in risk cases and every 6–12 months in affected patients | Annual in risk cases and every 6 months for affected patients | Annual between age 5 and 60 |
| AFG | Not routine | Not routine | Annual from 10 years onwards |
| Physical exploration and catecholamines in 24 h urine | Every 1–5 years from age 10 in risk cases and annually in affected patients | Annual | Annual |
| NMR/CAT brain and spinal chord | NMR from age 20. Every 10 years in risk cases and every 1–5 years for affected patients or with clinical suspicion | Baseline CAT basal in 1st–2nd decade and with focality and clinical suspicion | Every 3 years between age 15 and 40, then every 5 years from age 40 to 60 |
| Echography/abdominal CAT | Between age 15 and 20 every 1–5 years, echography and/or CAT | Abdominal echography. CAT is kidney carcinoma or pheochromocytoma is suspected | Every 3 years between age 20 and 60 (more frequently with multiple kidney cysts) |

**Table 2 – Diagnostic criteria for the von Hippel-Lindau disease.**

**With familial history**

*One or more of the following lesions are required:*

RCH

SNC hemangioblastoma

Organ lesions: kidney carcinoma, pheochromocytoma, renal/pancreatic cysts, pancreatic islet tumors, paragangliomas, epididymis cystadenomas, endolymphatic sac tumors

**Without familial history**

*RCH and/or SNC hemangioblastoma (if only one of these tumors expresses, a second organ lesion is necessary)*

## Clinical case

A male, 26, exhibited visual acuity reduction in the left eye with a 2-year evolution. The subject is a bearer of the mutation causing the VHL disease, father and brother with systemic syndrome expressions. Upon exploration, the VA of the LE is of 0.4. The LE ocular fundus exhibits orange-colored endophytic tumoration of two papilla diameters in the superior temporal arch with dilated nutrition vessels (Fig. 1). Optic coherence tomography (OCT) evidences a diffuse cystic edema in the posterior pole and angiography (AFG) reveals early hyperfluorescence of the lesion with delayed losses (Fig. 2). A systemic study was requested which in addition revealed solid nodules in both kidneys suggesting bilateral hypernephroma (subsequently confirmed after performing radical left nephrectomy), a hypercapturing nodule in the right supra-renal gland, a likely neuroendocrine pancreatic tumor and multiple

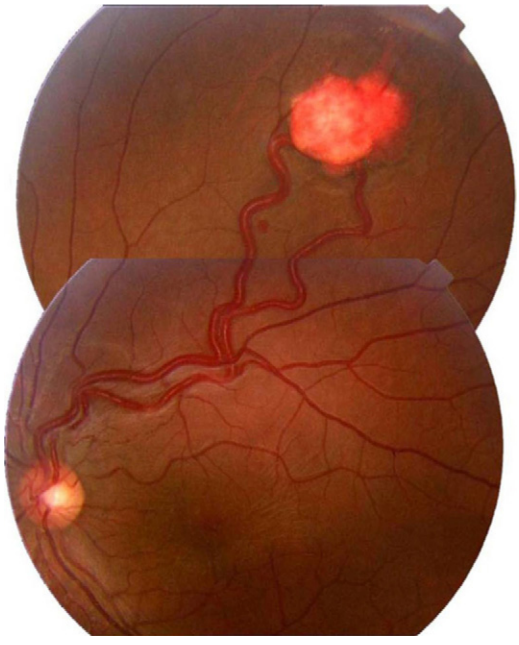


**Fig. 1 – Retinal capillary hemangioma.**