Retina

THE RARE DIAGNOSIS OF VON HIPPEL-LINDAU DISEASE IN A 29-YEAR OLD PATIENT

M.E. Sotirianakou, P. Bouzika, E. Papaconstantinou, I. Giachos, T. Paraskevopoulos,
Tryfon Rotsos

1st Department of Ophthalmology, University of Athens, Gennimatas General Hospital, Greece

Purpose: Von Hippel-Lindau disease (VHL) is a rare multisystem syndrome, characterized by multiple benign or malignant tumors and cysts in the retina, central nervous system and viscera; it is associated with a germline mutation in the VHL gene. The principal ophthalmic manifestation of VHL is retinal capillary hemangioblastomas (RCHs), which usually present with variable symptoms. This case report aims to describe the diagnostic procedure and treatment of bilateral asymmetrical RCHs.

Methods: A 29-year old white male presented to our hospital complaining of painless, progressive visual loss in his right eye. Visual acuity was 2/10 in the right eye and 10/10 in the left. Fundoscopy, optical coherence tomography (OCT) and fluorescein angiography (FA) were performed.

Results: Fundoscopy and OCT revealed 8 RCHs characteristic of VHL in the right eye, accompanied with extensive serous retinal detachment and macular exudates. In the left asymptomatic eye, there were 7 lesions, but the OCT images were unremarkable. The possible diagnosis of VHL prompted further imaging of the brain, abdomen and spine, which revealed kidney tumors. A genetic analysis confirmed the diagnosis of VHL. Treatment with laser photocoagulation was applied in all RCHs in both eyes. Ruthenium plaque radiotherapy was scheduled for 2 RCHs in the right eye due to size. Retinal detachment was completely resolved and there was subsequent improvement of the clinical manifestations. Furthermore, the visceral tumors present were managed surgically. At 3 months of follow-up, the patient maintains a good visual acuity of 9/10, waiting for brachytherapy in the right eye.

Conclusions: Our case emphasizes the role of the ophthalmologist in the diagnosis and care of VHL patients, as well as the need for individualized treatment.

Financial Disclosures: No