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Retinal capillary hemangioblastomatosis and renal tumor in Von Hippel-Lindau disease

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1. Case report

A 25 year-old man with no significant past medical history presented with 5 months of progressive vision loss in the right eye. Visual acuity was 20/400. Anterior segment exam was unremarkable. Fundoscopy (Fig. 1) and fluorescein angiography (Fig. 2) demonstrated multiple retinal capillary hemangioblastomas (RCHs) with associated exudative retinal detachment (RCHs), suggestive of von Hippel-Lindau (VHL) disease. Left eye exam was completely normal.

The patient was adopted with no known family history and denied any other systemic manifestations of VHL. A systemic workup was completed, which demonstrated a 2.5 × 2.0 × 2.6cm contrast-enhancing right renal mass (Fig. 3, arrow), concerning for renal cell carcinoma. The patient was referred to urology, endocrinology, and genetics. The patient received serial intravitreal bevacizumab injections in the right eye without improvement. Cryotherapy and vitrectomy were recommended, but the patient declined further treatment and unfortunately was lost to follow-up.

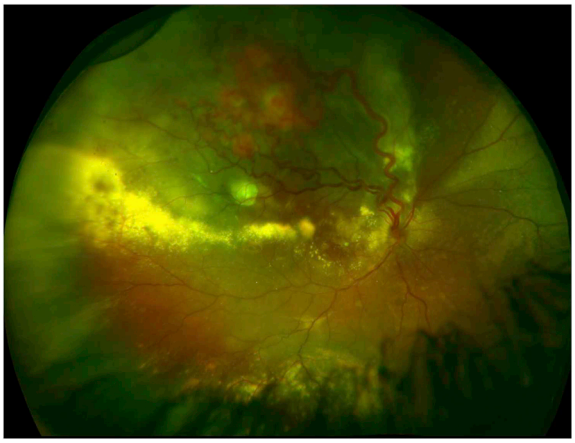

Fig. 1. Right Eye Fundus Photography. Multiple superotemporal retinal capillary hemangioblastomas with associated extensive exudation and retinal detachment involving the macula.

2. Discussion

VHL is an autosomal dominant, multisystem, progressive cancer syndrome caused by mutations of the VHL tumor suppressor gene, located on chromosome 3.[1] Clinical manifestations of VHL include central nervous system (CNS) and retinal capillary hemangioblastomas, renal tumors, pancreatic tumors, endolymphatic sac tumors, pheochromocytomas, and epididymal cystadenomas. 25–60% of patients with VHL will develop RCHs, which presents ophthalmologists a unique opportunity to diagnose a potentially life-threatening condition.[2] RCHs are hamartomatous retinal vascular tumors with distinct afferent and efferent feeder vessels as demonstrated in this case. Small lesions with

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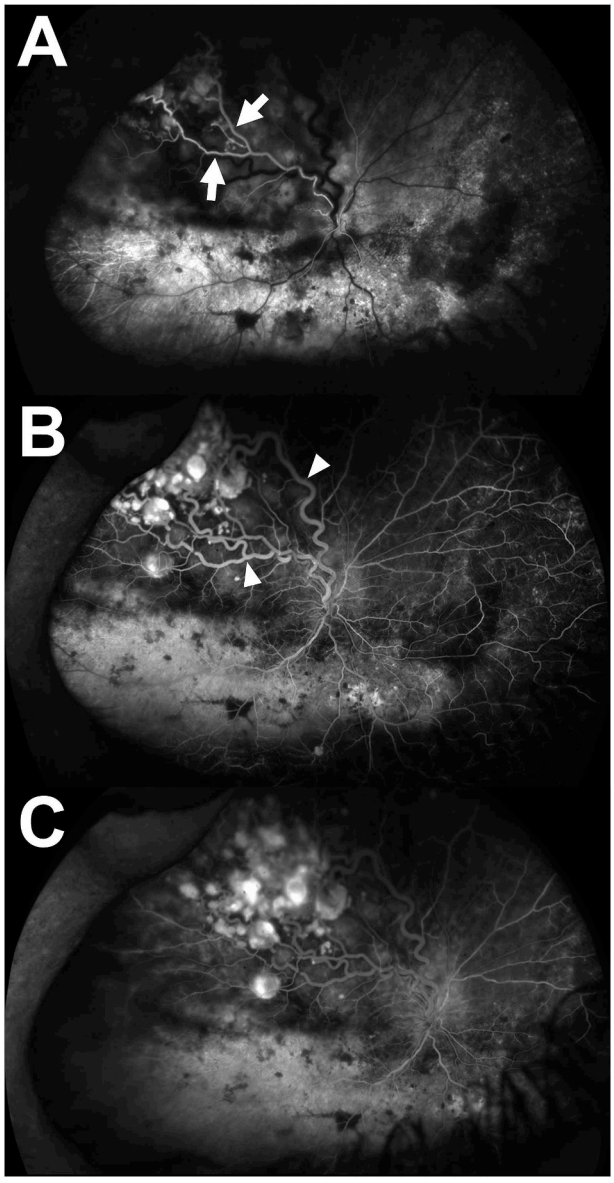

Fig. 2. Right Eye Fluorescein Angiography. (2A, 2B) Dilated and tortuous afferent (2A, arrows) and efferent (2B, arrowheads) feeder vessels are noted. (2C) Pooling of dye is noted in the retinal capillary hemangioblastomas.

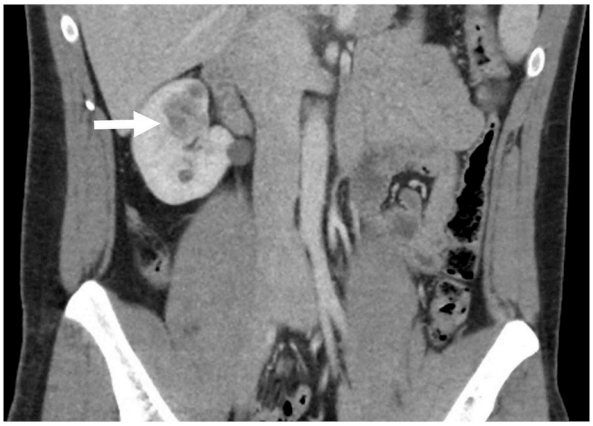

Fig. 3. Computed Tomography of the Abdomen and Pelvis. A contrast-enhancing right renal mass suspicious for renal cell carcinoma is noted (arrow).

limited exudation can be managed with laser or cryoablation, and anti-VEGF injections can be a useful adjunct; however, more extensive lesions with associated exudative or tractional retinal detachment may require vitrectomy.[3] Systemic screening should include i) magnetic resonance imaging (MRI) of the brain and spine (for CNS hemangioblastomas), ii) MRI abdomen (for renal cell carcinoma and pancreatic neuroendocrine tumors), and iii) plasma-free metanephrines or 24 hour urine fractionated metanephrines (for pheochromocytoma).[2] Patients with VHL require lifelong surveillance for tumor development.

3. Conclusions

Multiple RCHs are pathognomonic of VHL. Systemic workup and multidisciplinary surveillance should be pursued in all patients with VHL.

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Authorship

All authors attest that they meet the current ICMJE criteria for Authorship.

Consent

Written consent to publish this case has not been obtained. This report does not contain any personal identifying information.

Declaration of competing interest

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