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Case Report

Retinal capillary hemangioblastoma and hemiretinal vein occlusion in a patient with primary congenital glaucoma: A case report

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Abstract

The presence of retinal capillary hemangioblastoma and cerebellar hemangioblastoma in the context of Von Hippel-Lindau syndrome (VHL) is not characteristically associated with other ophthalmologic conditions. Here, we report the case of a 22-yearold female with a history of bilateral primary congenital glaucoma who presented with a right juxtapapillary retinal capillary hemangioblastoma and an old hemiretinal vein occlusion in which the retinal capillary hemangioblastoma was likely the contributing factor. Her systemic work up was positive for VHL syndrome and revealed the presence of a fatal large brainstem hemangioblastoma. To our knowledge, the association of VHL and congenital glaucoma and/or retinal venous occlusion has not been reported.

Keywords: Capillary hemangioblastoma, Von Hippel Lindau syndrome, Hemiretinal vein occlusion, Primary congenital glaucoma

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Introduction

Retinal capillary hemangioblastomas (RCHs) are vascular hamartomas that can arise at any part of the retina. They may occur sporadically or be part of the autosomal dominantly-inherited condition known as Von Hippel-Lindau (VHL) syndrome.[1]

VHL syndrome is considered a systemic cancer disorder. RCHs are often its most common and earliest presentation and the presence of multiple or bilateral RCHs is highly suggestive of VHL syndrome. The RCH typically presents as a solitary ''vascular'' tumor in the superior temporal region of the peripheral retina in a patient in the mid-twenties. The tumor, or tumors, generally start small but tend to grow rapidly and lead to many ocular complications including excessive lipid exudation, exudative retinal detachment, traction retinal detachment and neovascular glaucoma.[2] Submacular fluid and macular edema are common complications when the RCH occurs in the juxtapapillary region.[2] In addition to the retinal location, hemangioblastomas may also occur at the optic nerve in patients with VHL, and must to be differentiated from optic disc edema, papillitis, sarcoidosis, or anterior ischemic optic neuropathy.

In addition to the ocular manifestations, the spectrum of VHL syndrome also includes central nervous system (CNS) tumors. Hemangioblstomas at the cerebellum and spinal cord are the most common site of CNS involvement. These tumors, primarily at the cerebellum, have been described in about 5% of the patients with ocular involvement. Pancreatic cancers and cysts, pheochromocytomas[3] and renal cell

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carcinomas are also common. The latter is the leading cause of death in the VHL syndrome. To the best of our knowledge the RCHs in VHL have not been reported in association with retinal venous occlusive disease or congenital glaucoma. We here report a 22-year-old female patient with a history of congenital glaucoma since birth, in whom hemiretinal vein occlusion and retinal and CNS hemangioblastomas were discovered incidentally and in where the RCH may have contributed to the retinal venous occlusion.

Case report

A 22-year-old female was referred to the retina clinic for evaluation of a juxtapapillary lesion in the right fundus. Her past ocular history disclosed bilateral childhood glaucoma discovered soon after birth for which the patient underwent multiple bilateral glaucoma surgeries. On the day of examination her vital signs were stable. Snellen visual acuity was 20/300 in the right eye and only perception of light in the left eye. Intraocular pressure was 16 mmHg for the right eye and 14 mmHg for left eye without glaucoma medication. Slit lamp examination showed a clear cornea and clear lens in the right eye, but a large opaque cornea and pseudophakia in the left eye. Dilated fundus exam of the right eye showed advanced optic disc cupping, a superonasal juxtapapillary reddish retinal vascular mass of approximately half disc area in size, surrounded by a faint ring of retinal exudates suggestive of a RCH [Fig. 1]. The mass was located at the level of the upper trunk of the central retinal vein as it exited the optic disc hiding the visualization of this venous trunk. The branches of this upper trunk however, displayed, in both the temporal and nasal periphery, venous tortuosity and collateral channels throughout the upper half of the retina which connected to the inferior retinal venous circulation. Furthermore, sclerosed veins were seen at the superior arcade [Fig. 1]. All these signs pointed to an old superior hemiretinal vein occlusion. Examination of the left fundus was limited in visibility by the corneal opacification, but no posterior pole pathology was detected on B-scan ultrasonography.

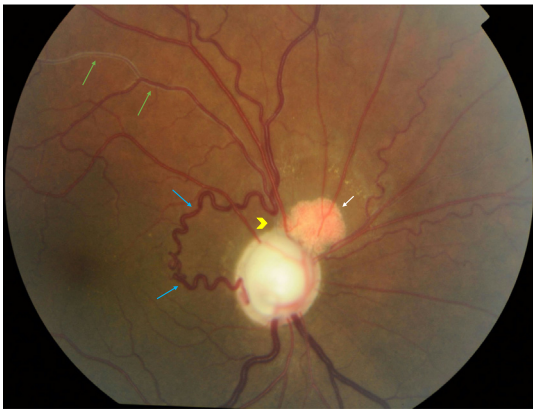

Fig. 1. Color fundus photograph of the right eye. Visible on the photograph are advanced optic disc cupping and a superonasal juxtapapillary reddish retinal vascular mass of approximately half disc area in size (white arrow), surrounded by a faint ring of retinal exudates suggestive of a retinal capillary hemangioblastoma. A longstanding superior retinal vein occlusion is also present demonstrated by (1) sheathing of the retinal vein at the superotemporal aracade (green arrows), (2) marked stenosis of the proximal segment of the superior retinal vein adjacent to the mass (yellow arrowhead), and (3) the establishment of venous collateral channels draining the superior venous blood towards the ciliary circulation (blue arrows).

Spectral domain optical coherence tomography (SD-OCT) of the right fundus demonstrated that the juxtapapillary mass was hyper-reflective and intraretinal in location [Fig. 2A and B]. The surrounding neurosensory retina displayed intraretinal hyporeflective spaces and retinal thickening corresponding to the clinically visible retinal edema [Fig. 2C and D]. There was no macular edema.

Fundus fluorescein angiography (FFA) of the right eye showed early filling of the juxtapapillary lesion with leakage of dye in the later stages of the study. In addition, delayed filling of the superior retinal venous system with normal filling time of the inferior retinal veins was noted as well as the presence of many retinal venous collaterals connecting the veins of the superior half of the retina to the inferior ones, all of which were indicative of a longstanding superior hemiretinal vein occlusion [Fig. 3A, B, C]. No retinal capillary dropout could be detected. The FFA of the left eye showed, despite a hazy view, the presence of a hyperfluorescent late-leaking lesion at the temporal part of the macula which was highly suggestive of a RCH [Fig. 3 D]. With features of bilateral RCHs, the diagnosis of VHL was highly suspected and additional family history was taken. It revealed no parental consanguinity. Additionally, the patient's mother and grandmother had died at an early age from brain tumors. The patient's 28-year-old sister, who had no ocular symptoms, disclosed that she had multiple spinal cystic lesions and multiple large pancreatic cysts. Her ocular examination revealed peripheral RCH on her right eye.

The patient was referred for systemic evaluation. The basic laboratory work-up was within normal limits. Magnetic resonance imaging (MRI) of the brain with intravenous gadolinium contrast demonstrated a well-defined highly vascular mass inside the brainstem with surrounding edema representing a large hemangioblastoma. However, there were no apparent neurological or behavioral deficits upon her evaluation in the retina clinic. The patient was referred to a general hospital for urgent neurosurgical consultation. Excision of the brainstem hemangioblastoma was decided and the patient underwent neurosurgery. Unfortunately, following the surgery she entered into a deep coma and passed away.

Discussion

Retinal capillary hemangioblastomas (RCHs) classically start as small, sometimes pinpoint, red or grey retinal lesions. As the vascular tumor grow, they become larger and more globular in appearance.[4] RCHs commonly occur as peripheral tumors in the retina, most frequently in the superotemporal or inferotemporal quadrants and are associated with prominent feeder vessels. In 11–15% of cases they occur in a juxtapapillary location.[5] Complications of RCHs are commonly macular or extramacular fluid and lipid exudation, epimacular membrane formation, exudative retinal detachment, tractional retinal detachment, vitreous hemorrhage and neovascular glaucoma.

The case presented herein is interesting in that a RCH in a juxtapapillary location was associated with superior hemiretinal vein occlusion (HRVO). Although retinal vein occlusion, with or without central retinal artery occlusion, has been reported in other juxtapapillary lesions such as optic disc

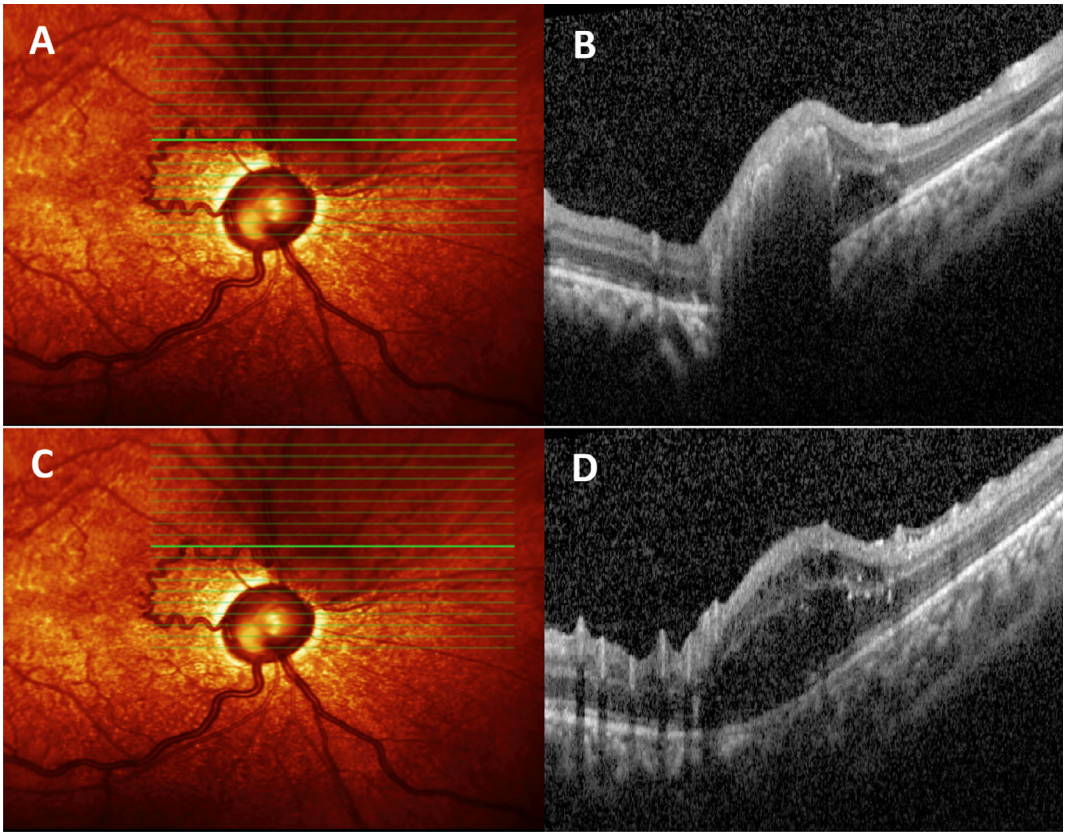

Fig. 2. Spectral domain optical coherence tomography (SD-OCT) of the right fundus. (A and B) The SD-OCT line cut seen in (B) corresponds to the green line of the raster scan seen in (A) and demonstrates the juxtapapillary hyper-reflective intraretinal mass with posterior shadowing. (C and D) The SD-OCT line cut seen in (D) corresponds to the green line of the raster scan immediately superior to the tumor seen in (C) and demonstrates intraretinal hypo-reflective spaces and thickening of the neurosensory retina corresponding to the clinically visible retinal edema.

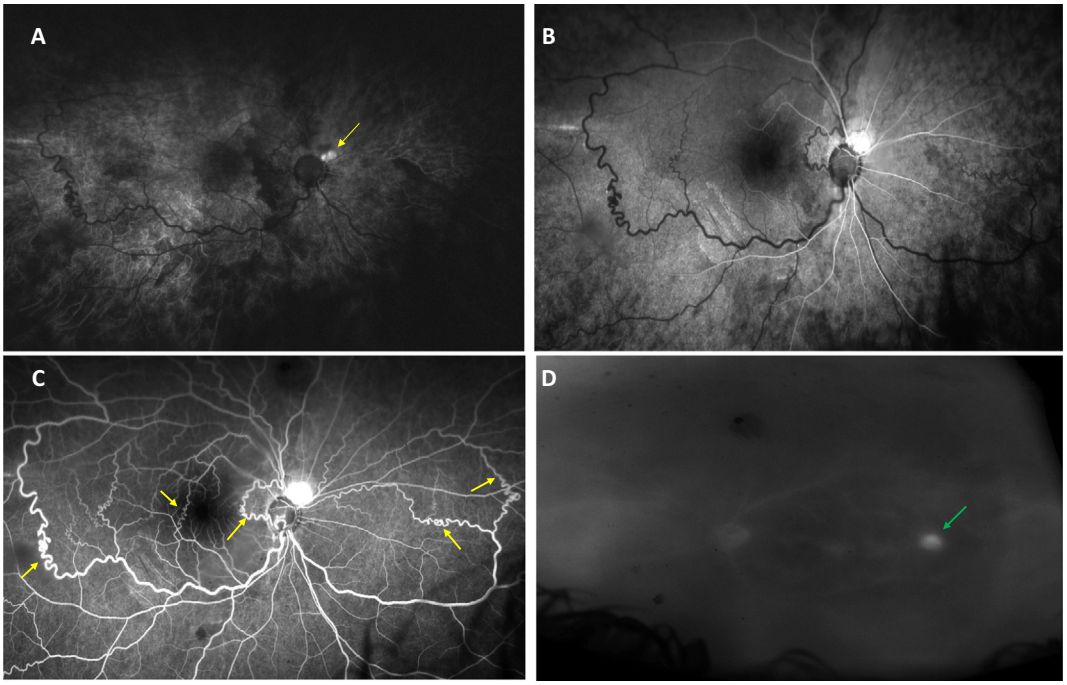

Fig. 3. Fundus fluorescein angiography (FFA). FFA of the right eye at 12 s showing early filling of the juxtapapillary lesion (yellow arrow). FFA of the right eye at 15 s showing increased hyperfluorescence of the juxtapapillary lesion. FFA of the right eye at 27 s demonstrating increased leakage of fluorescein from the juxtapapillary lesion and numerous retinal venous collaterals, all of which cross the horizontal raphe (yellow arrows), draining the superior retinal venous blood into the inferior venous system. FFA of the left eye at 59 s showing a hyperfluorescent late-leaking lesion at the temporal part of the macula suggestive of a retinal capillary hemangioblastoma (green arrow).

melanocytoma[6] to our knowledge, the association of RCH and HRVO has not been reported. The exact mechanism of retinal venous occlusion occurring in association with juxtapapillary lesions has not been fully elucidated. We hypothesize that in our case the location of this growing lesion over the superonasal edge of optic disc probably contributed to a chronic compression of the superior trunk of the hemiretinal vein which consequently lead to a longstanding slowing of the blood flow drained through this vein resulting in a chronic superior HRVO picture. As a consequence of this chronic occlusion, many retinal venous collateral channels have developed in this case to drain the superior hemiretinal blood into the inferior hemiretina. No demonstrable retinal ischemia was noted in this case.

We believe that the HRVO in our case most likely occurred progressively and not as a sudden thrombotic event. Firstly, there was no history of sudden visual loss. Secondly, no significant capillary non-perfusion could be demonstrated throughout the retina on fundus fluorescein angiography. This finding in association with insidious development of significant retinal venous collaterals leads us to consider that HRVO occurred as a result of a slow compressive mechanism. However, since the patient also exhibited severe visual loss secondary to advanced glaucoma, an acute but unrecognized sudden HRVO cannot be totally excluded. Furthermore, the presence of glaucoma itself which is a known risk factors for retinal vein occlusion may also have been the primary mechanism for the HRVO.[7]

Our case also represents an unusual ophthalmic association. We are unaware of previous reports describing VHL associated with primary congenital glaucoma. Congenital glaucoma is common and often inherited in an autosomal recessive pattern in the Saudi population.[8] Our patient was the only member of her family who had congenital glaucoma. Whether the combination of congenital glaucoma and VHL in our case represents a simple association of two independent hereditary diseases or the development of a new mutation in VHL gene is yet unknown.

Hemangioblastomas involving CNS are known to be benign tumors and their recurrence rate is 15% to 25% after surgical removal.[9] They rarely behave aggressively. In our patient, there was a strong family history of death at an early age from brain tumors in the patient's mother and grandmother. This indicates the presence of an aggressive type of VHL in this pedigree. Early detection of these tumors associated with VHL and proper management are crucial. It is recommended for patients with VHL to undergo annual complete physical examination and dilated eye examinations, renal ultrasonography, and 24-hour urine collection for vanillylmandelic acids. Affected patients should undergo neuroimaging every 3 years up to the age of 40 years and every 5 years thereafter.[10] Relatives at-risk should also undergo thorough annual screening for the disorder. RCHs often present before other systemic tumors become apparent and the ophthalmologist may be the first person to diagnose this potentially fatal disease.

Conclusions

We present a case of juxtapapillary retinal capillary hemangioblastoma complicated by hemiretinal vein occlusion in patient with congenital glaucoma and Von Hippel Lindau syndrome. Furthermore, a strong history of lethal central nervous system hemangioblastomas was noted in our patient and her family. To our knowledge, the association of VHL and congenital glaucoma and/or retinal venous occlusion has not been reported.

Conflict of interest

The authors declared that there is no conflict of interest.

Acknowledgement

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