Retinal capillary hemangioblastoma associated with retinochoroidal coloboma in Von Hippel-Lindau disease

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Case Report

A 30-year-old lady of diagnosed von Hippel-Lindau (VHL) disease was referred to us for ophthalmological evaluation. Her visual acuity was 6/6 in both eyes and anterior segment examination was normal. On fundus examination, bilateral multiple retinal capillary hemangioblastomas (RCH) were seen within and around the inferior retinochoroidal colobomas [Figs. 1-4]. Multiple feeder arteries and tortuous, beaded draining veins were seen. Fundus fluorescein angiography showed leakage [Fig. 5]. One sitting of barrage laser was done around the coloboma. Patient did not consent to further treatment as there was no ocular symptom. At 1 year of follow-up although slight increase in size of some tumors were noted, visual acuity remained stable and no complications were seen.

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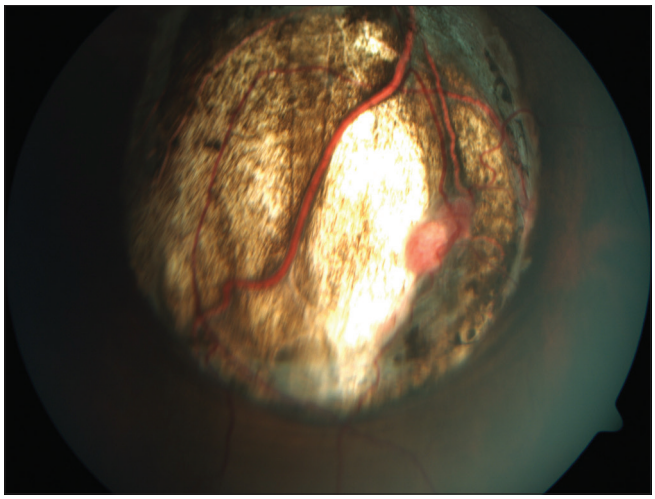

Figure 1: Color fundus photograph of right eye showing multiple capillary hemangioblastomas aggregated over inferior retinochoroidal coloboma

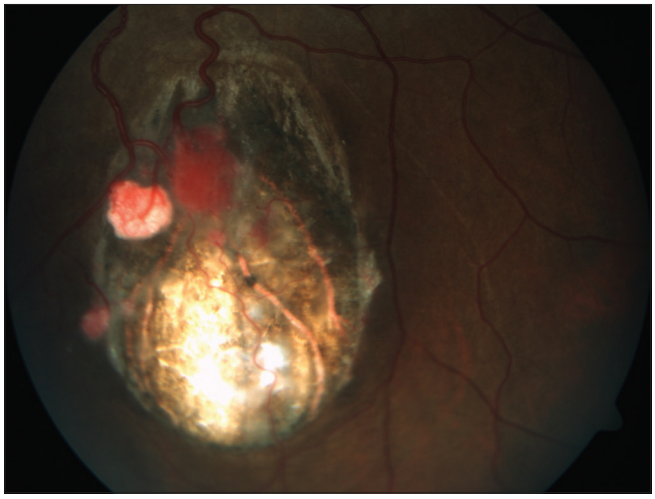

Figure 3: Color fundus photograph of left eye showing multiple capillary hemangioblastomas aggregated over inferior retinochoroidal coloboma

Discussion

VHL disease is an autosomal dominant disorder characterized by tumors and cysts in multiple organs due to mutation in VHL tumor suppressor gene on chromosome 3. RCH is one of the most frequent presenting feature of the disease occurring in 49% to 85% cases.[1] Retinochoroidal coloboma is a rare eye malformation, can be sporadic or inherited, frequently with mutation in *PAX2* gene, and is known to be part of various genetic syndromes. Association of retinochoroidal coloboma with VHL disease or RCH with coloboma in VHL disease has never been reported.

Capillary hemangioblastoma can cause vision loss from exudation, haemorrhage, and retinal detachment. The most common complication associated with retinochoroidal colobomas is retinal detachment, occurring in 8.1–43% of cases.[2,3] Presence of both entities, as in our case, can increase the risk by several folds.

Lasave and Deromedis reported bilateral chorioretinal coloboma and a solitary, unilateral RCH and suggested a possible correlation between ischemic areas of the coloboma and production of vascular endothelial growth factor

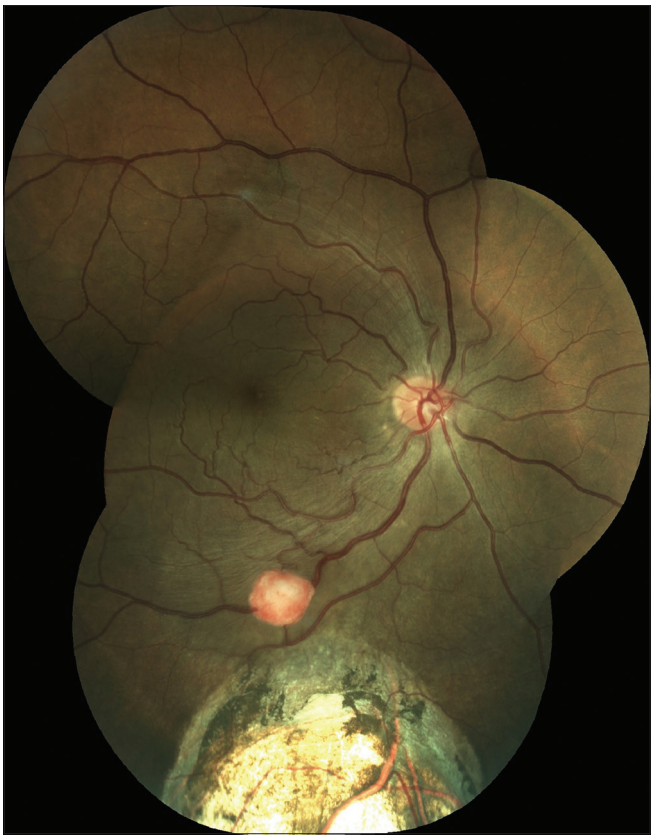

Figure 2: Color fundus montage of right eye showing one capillary hemangioblastomas just above inferior retinochoroidal coloboma

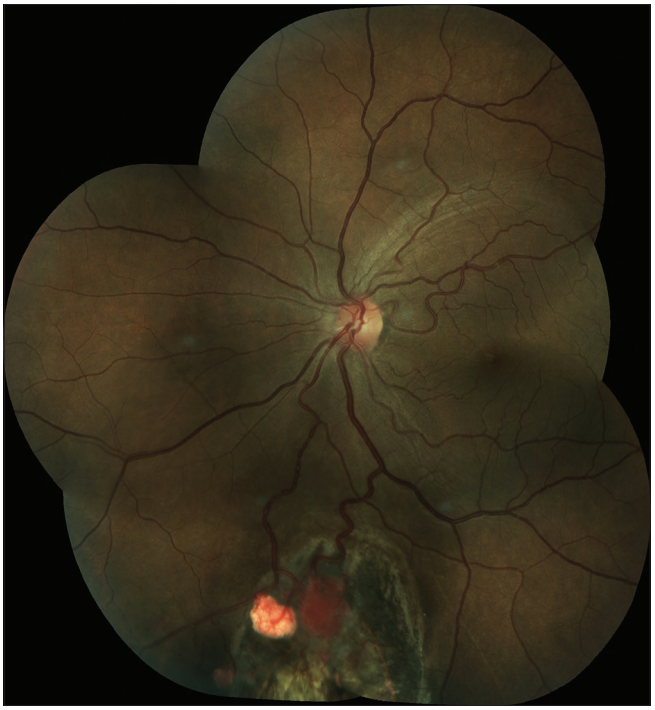

Figure 4: Color fundus montage of left eye of the same patient showing inferior coloboma along with multiple capillary hemangioblastomas

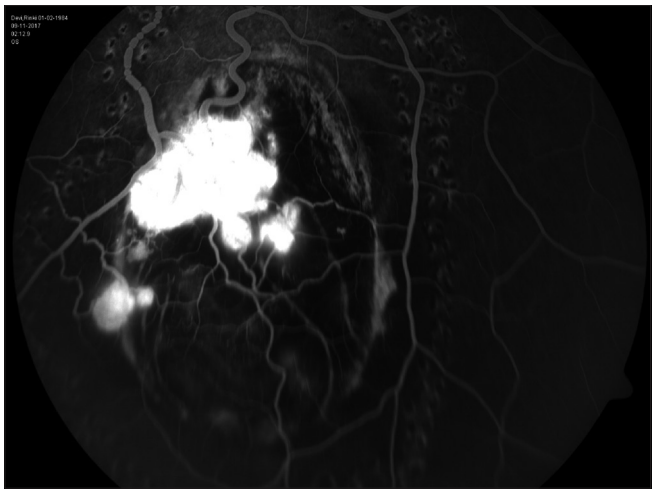

Figure 5: Fundus fluorescein angiography picture of left eye showing leakage from all capillary hemangioblastomas

leading to abnormal vessel growth in the form of retinal hemangioblastomas.[4] In our case, bilateral aggregation of the retinal hemangioblastomas over and around the coloboma adds to the above hypothesis that some factors like ultrastructural abnormality of the colobomatous area or ischemia definitely play a role in this association rather than being a mere coincidence. Further genetic studies will help to establish the possible syndromic association between these two entities.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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