# A 48-Year-Old Male with a Retinal Vascular Anomaly

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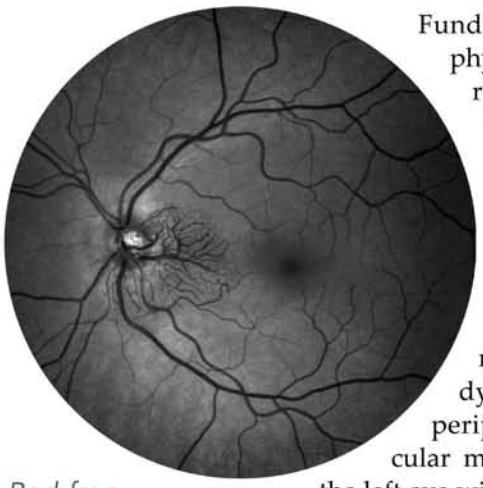
A 48 year-old male was referred to The Retina Institute for evaluation of retinal vascular changes in his left eye. The patient had no visual changes or complaints and denied any past ocular or medical history. His family history consisted of unspecified cancer in his father and maternal grandmother, and a review of systems was positive for frequent epistaxis.

Visual acuity measured 20/20 in both eyes. There was no relative afferent pupillary defect. Intraocular pressure by tonometry was 18 OU. Anterior segment exam was significant for mild vascular tortuosity of conjunctival vessels and mild cataracts in both eyes.

Dilated fundus exam revealed clear vitreous, sharp and pink optic nerves, mild macular RPE mottling, and normal periphery in both eyes. The vasculature was normal in the right eye but a telangiectatic, branching vascular anomaly concentrated along the papillomacular bundle was found in the left eye (Figure 1 - featured on the September 2014 cover of *Ophthalmology, Volume 121, Number 9*).



*Red free*

Fundus photography and a fluorescein angiogram (FA) were obtained. The FA was normal in the right eye but revealed rapid transit of dye through a peripapillary vascular malformation in the left eye without leakage

Upon further questioning, the patient did endorse a family history of hemorrhagic telangiectasias. Therefore, the patient was diagnosed with a retinal vasculature anomaly from hereditary hemorrhagic telangiectasia (HHT, aka Osler-Weber-Rendu disease).

## Diagnosis:

The differential diagnosis for this patient's retinal vascular anomalies from HHT includes peripapillary choroidal neovascular membrane (degenerative, inflammatory/infectious, idiopathic causes), neovascularization of the disc (proliferative diabetic retinopathy, retinal vein occlusion, arterial insufficiency), prepapillary vascular loop, or Bergmeister's papilla. The lack of leakage on FA ruled out a choroidal neovascular membrane and neovascularization of the disc. Furthermore, the extensive branching vascular network would be uncharacteristic of prepapillary vascular loops and a Bergmeister's papilla.

*Figure #1*

HHT is a rare autosomal dominant disorder characterized by abnormal mucocutaneous telangiectasias and arteriovenous malformations (AVMs) which have a tendency to bleed. These vascular abnormalities most commonly occur in the skin, oral cavity, nasopharynx, eye, central nervous system, lungs, and abdominal viscera. The reported annual incidence is 1-2 cases per 100,000 population with an overall prevalence of approximately 1-2 cases per 10,000 population. It occurs with equal frequency in males/females and is most common among whites[1].

## Ocular Involvement:

Due to the rare nature of HHT, most literature sources involve retrospective case series which report ocular involvement in 45-65% of patients[2]. The most common ophthalmic finding is conjunctival telangiectasias with hematic epiphora (32-65%). "Mulberry-like" iris vascular malformations at the pupillary margin have been associated with spontaneous hyphema[3].

Vascular abnormalities of the posterior segment have been identified in up to 37% of reported case series[4]. The first description was published by Francois in 1938 and subsequent literature have described a variety of retinal findings including vessel tortuosity/dilation, telangiectasias, arteriovenous malformations, fusiform aneurysms, perivascular hemorrhage/exudates, neovascularization, artery occlusions, and ectatic optic nerve vessels (as seen in our patient)[5-7]. Vascular abnormalities may present diffusely throughout the retina or may be located in the macula leading to vision loss[3,8].

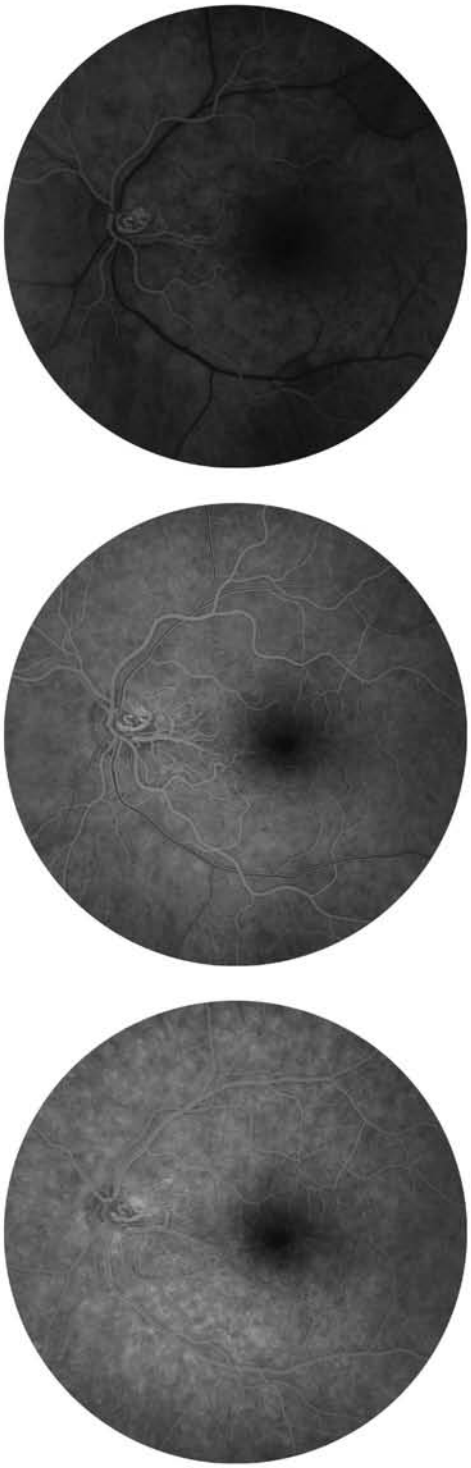
Choroidal changes have also been noted with some patients exhibiting choriocapillaris atrophy or choroidal telangiectasias in the form of prominent choroidal vessels with associated serous retinal pigment epithelial detachments which resolved spontaneously[4,9]. Nonsimultaneous choroidal hemorrhages has been reported intraoperatively during a vitrectomy for one eye and after an uncomplicated cataract surgery in the other eye[10].

## Ocular Treatments:

Symptomatic macular telangiectasias have been treated with photodynamic therapy and transconjunctival cryotherapy and photocoagulation have been used for recurrent vitreous hemorrhage and neovascularization[6,8]. Although anti-VEGF therapy would likely help manage complications from abnormal vasculature, no use of anti-VEGF agents has been reported in literature yet.

## Systemic Manifestations:

HHT affects many parts of the body including the skin, oral cavity, nasopharynx, heart, lung, liver, gastrointestinal tract, and central nervous system. Patients commonly present with recurrent epistaxis episodes and skin telangiectasia. More serious ailments include dyspnea on exertion, gastrointestinal bleeding, and strokes from hemorrhage or arteriovenous shunting through abnormal vessels.



*Early, laminar, late FA phases*

Symptom onset is often delayed until the fourth decade of life with about 90% of patients manifesting by age 40[1]. The prognosis varies, depending on the severity of symptoms. With appropriate screening and aggressive medical/surgical management of bleeding, life expectancy for the majority of patients may approach that of the normal population.

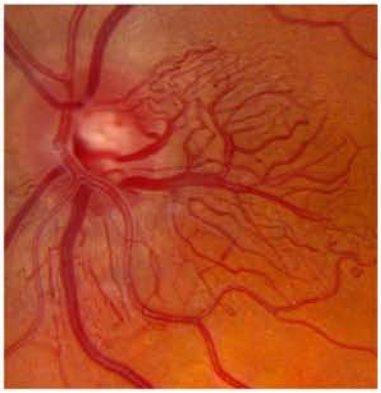
## Systemic Work-up:

Because of the presence of AVMs and associated sequelae, patient evaluations with a primary care physician should include a thorough physical exam, complete blood count, coagulation profile to exclude concurrent coagulopathy, urinalysis to screen for hematuria, hemoccult test, and liver function tests. Further work-up may include computed tomography and magnetic resonance imaging techniques to screen for cerebral, pulmonary, or abdominal visceral AVMs; cardiac echocardiography for intracardiac shunts; and upper/lower endoscopy for GI bleeding.

HHT is a clinical diagnosis and the likelihood of affliction is based on the presence of recurrent epistaxis, mucocutaneous telangiectasias, visceral AVMs, and first-degree relative with HHT (Curacao criteria). Skin biopsy findings may aid in diagnosis and genetic testing can confirm the presence of mutations within endoglin, activin receptor-like kinase type I, and SMAD4 genes.

## Conclusion:

HHT is a rare autosomal dominant disorder characterized by abnormal mucocutaneous telangiectasias and arteriovenous malformations throughout the body which have a tendency to bleed. Conjunctival telangiectasias is the most common ocular manifestation but vascular malformations of the posterior segment have been reported in up to 37% of reported case series.



*Detail of anomaly*

While these vascular changes often do not affect vision, the treatment of symptomatic telangiectatic vessels and neovascularization has not been well-described yet. The complications of HHT are potentially serious and can not only lead to vision loss but can also lead to heavy bleeding, organ dysfunction, stroke, and even death. Close follow-up in conjunction with a primary care physician is vital to identify and aggressively manage sequelae.

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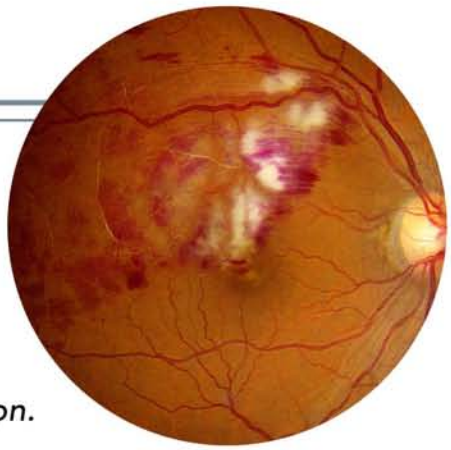

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