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Hereditary hemorrhagic telangiectasia with multiple fusiform retinal aneurysms

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Introduction

Hereditary hemorrhagic telangiectasia (HHT) is a rare genetic disorder characterized by abnormalities of vascular structures involving skin, mucosal membranes, and other organs. A wide variety of clinical manifestations have been associated with HHT ranging from small telangiectasia on face causing only cosmetic problems to life-threatening visceral arteriovenous malformations leading to systemic emboli, hepatic dysfunction, and high output cardiac failure. These diverse clinical manifestations develop according to the localization and the severity of the vascular abnormalities.

Ocular involvement is common in HHT and conjunctival telangiectasia is the most common ocular lesion. Retinal telangiectasia, retinal arteriovenous malformations, and choroidal telangiectasia are other relatively rare ocular findings [1–3]. We herein present a case of HHT with bilateral multiple retinal fusiform aneurysms and conjunctival telangiectasia.

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Case report

A 38-year-old woman with the diagnosis of HHT was examined from the dermatology clinic for investigation of ocular involvement. She had a history of recurrent epistaxis and there were several spots of telangiectasia distributed on her arm, back, thigh, and buccal mucosa especially apparent for the last 3 years (Figs. 1 and 2). Her family history was negative for HHT and she was not taking any anticoagulant medication.

The best corrected visual acuity was 10/10 in both eyes. Numerous conjunctival telangiectasias on nasal bulbar conjunctiva and spider-like angiomatous malformations on the caruncula and the forniceal conjunctiva in the neighborhood of the caruncula were noted during biomicroscopic examination (Fig. 3). Fundus examination revealed tortuosity and multiple fusiform dilatations in all of the branches of the retinal vein and fine telangiectatic capillaries in both eyes (Fig. 4). The macula and the optic disc were normal. Neither hemorrhage nor exudate was observed. The fundus fluorescein angiogram did not show any leakage or capillary nonperfusion while the walls of retinal veins were stained (Fig. 5). The OCT examination of the macula was normal. The investigations done for the patient included: kidney and liver function tests including HbA1c and spot glucose level, ACE level, chest radiography, erythrocyte sedimentation rate, HLA B_{51}, rheumatic factor, antinuclear antibodies, double-stranded DNA, antineutrophil cytoplasmic antibodies, C-reactive protein, complement level (C3 and C4), *Brucella*, toxoplasmosis, and *Borrelia* titers, HBsAg, VDRL level, prothrombin time, partial thromboplastin time, and hemoglobin electrophoresis. Of these, only HLA B_{51} was positive and all other results were normal. Pathological

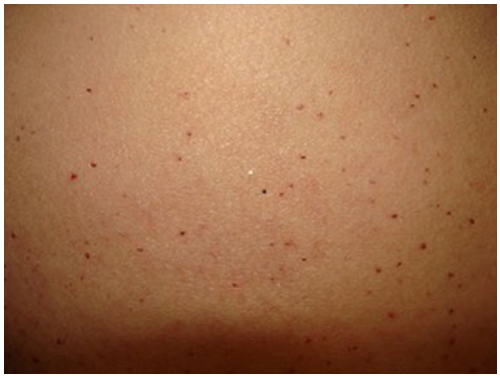

Fig. 1 Widespread telangiectatic lesions on her back

examination of biopsy material taken from lesions in the back region showed ectatic proliferating small vascular structures in the papillary dermis. Since only one minor criterion, HLA B_{51} positivity, was present and any other major findings were absent, the diagnosis of Behçet's disease was not being considered.

Discussion

The ophthalmological findings observed in this patient were conjunctival and retinal vascular abnormalities. The clinical and the angiographic appearances of retinal lesions were characterized by increased tortuosity and multiple aneurysmal dilatations along retinal veins without associated findings such as exudate, hemorrhage, leakage, or capillary nonperfusion. As extraocular findings, she had multiple mucocutaneous telangiectasia involving her back, extremities, inguinal area, and buccal mucosa. The patient was complaining of frequent epistaxis also.

Generalized mucocutaneous telangiectasia with ocular involvement is seen in very few conditions. Mainly, generalized essential telangiectasia (GET), ataxia telangiectasia (AT) and HHT are the diseases where concomitant ocular and cutaneous telangiectasias were reported.

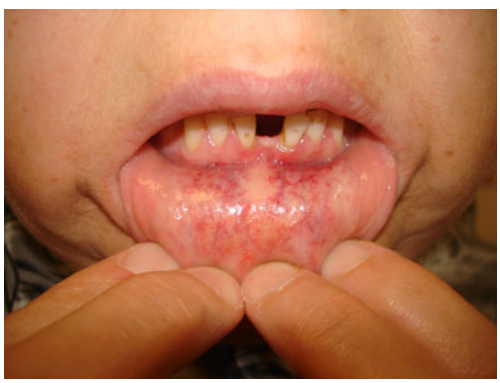

Fig. 2 Telangiectasia on her buccal mucosa

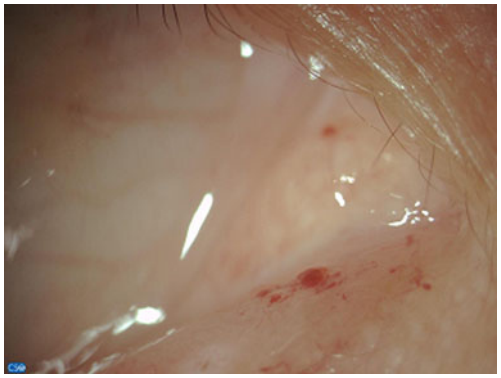

Fig. 3 Multiple telangiectatic vessels and spider-like angiomatous malformations on the caruncula

Generalized essential telangiectasia is a rare disease affecting young healthy women. The condition is characterized by spontaneous development and progressive spreading of telangiectasia. Although the age and the clinical course of skin lesions of our patient may resemble to GET, the presence of epistaxis, which is not the component of GET, excluded the diagnosis. Moreover, although conjunctival involvement was reported, retinal involvement also has not been reported with GET.

Ataxia telangiectasia is another disease in which accompanying cutaneous and ocular telangiectasias are seen. In this disease, the age of onset is the first 2 years of life with progressive neurological dysfunction, immunodeficiency, and recurrent sinopulmonary infections. The age and general health status of our patient can easily exclude the diagnosis of AT. Retinal telangiectasia also is not a reported finding of AT.

Idiopathic retinitis, vasculitis, aneurysms, and neuroretinitis (IRVAN) syndrome is a rare characterized by the presence of multiple aneurysmal dilatations along the arteriolar tree. Three major criteria of IRVAN are retinal vasculitis, aneurysmal dilatations at arterial bifurcations, and neuroretinitis which do not conform to the findings of our patient [4]. Moreover, IRVAN is limited to eye and extraocular findings

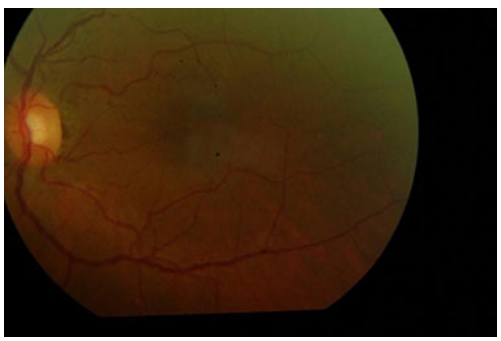

Fig. 4 Increased tortuosity and multiple fusiform aneurysms along the inferior and superior temporal retinal veins and fine telangiectatic capillaries temporal to macula

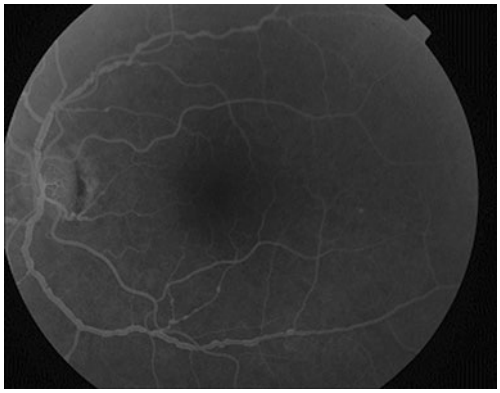

Fig. 5 Angiographic images of the patient with no hemorrhage or exudate. Retinal vessel walls were stained with fluorescein

of our patient such as epistaxis and mucocutaneous telangiectasia are not found in IRVAN.

Hereditary hemorrhagic telangiectasia is also another disorder in which both retinal and conjunctival telangiectasia with mucocutaneous telangiectasia is seen. HHT is a rare vascular disease characterized by vascular abnormalities. The hallmark of vascular lesions in HHT is the thinning and the dilatation of the vessel walls which make the vessels friable and increase the risk of bleeding. Whole vasculature throughout the body may be affected and may lead to the manifestations such as epistaxis, gastrointestinal bleedings, mucocutaneous telangiectasia, and pulmonary embolism.

Ocular involvement is common (45–65 %) in HHT and the most frequent ocular location is conjunctival telangiectasia. It has been reported in 35–38 % of HHT cases and it is harmless being not accepted as an indicator of visceral involvement [1–3].

In contrast to conjunctiva, intraocular involvement of HHT is a rare finding and includes abnormalities in the retinal and the choroidal vascular beds. Retinal vascular abnormalities, which had been reported in 10 % of cases, are the tortuosity and the dilatation of the retinal vessels, retinal telangiectasia, new optic disc vessels, and retinal arteriovenous malformations [1, 3, 5] while the only reported choroidal lesion which may lead to surgical complications is telangiectasia [6].

Landau et al. reported a family of HHT with both conjunctival and retinal telangiectasia and described the retinal lesions as tortuous veins resembling twisted cords and suggested varices [5]. Brand observed retinal involvement in 2 of 20 HHT patients. One of them had numerous telangiectatic vessels on papillomacular bundle and other had multiple foci of fine telangiectasia throughout the retina [1]. Ocular lesions in HHT are generally stable and rarely cause symptoms. Only in one case with symptomatic parafoveal telangiectasia photodynamic therapy had been reported [7]. To our knowledge, multiple fusiform aneurysms had never been reported before as a finding of HHT.

Conflict of interest None of the authors has conflict of interest with the submission.

References

1. Brant AM, Schachat AP, White RI. Ocular manifestations in hereditary hemorrhagic telangiectasia (Rendu-Osler-Weber disease). Am J Ophthalmol. 1989;107:642–6.
2. Geisthoff UW, Hille K, Ruprecht KW, et al. Prevalence of ocular manifestations in hereditary hemorrhagic telangiectasia. Graefes Arch Clin Exp Ophthalmol. 2007;245:1141–4.
3. Vase I, Vase P. Ocular lesions in hereditary haemorrhagic telangiectasia. Acta Ophthalmol (Copenh). 1979;57:1084–90.
4. Samuel MA, Equi RA, Chang TS, et al. Idiopathic retinitis, vasculitis, aneurysms, and neuroretinitis (IRVAN): new observations and a proposed staging system. Ophthalmology. 2007;114:1526–9.
5. Landau J, Nelken E, Davis E. Hereditary haemorrhagic telangiectasia; with retinal and conjunctival lesions. Lancet. 1956;271:230–1.
6. Mahmoud TH, Deramo VA, Kim T, et al. Intraoperative choroidal hemorrhage in the Osler-Rendu-Weber syndrome. Am J Ophthalmol. 2002;133:282–4.
7. Mennel S, Hoerle S, Meyer CH. Photodynamic therapy in symptomatic parafoveal telangiectasia secondary to Osler-Rendu-Weber disease. Acta Ophthalmol Scand. 2006;84:273–5.