Retinal vascular abnormalities in Sturge-Weber syndrome

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Sturge–Weber syndrome (SWS) includes facial, leptomeningeal and choroidal hemangioma. The retinal vasculature is essentially normal. Rare cases of retinal vascular tortuosity and arterio-venous malformations have been reported. We report two cases with rare concomitant retinal vascular abnormalities along with SWS. Both the patients had nevus flammeus, hemifacial hypertrophy, and choroidal hemangioma. In one case, retinal cavernous hemangioma was seen in the affected eye. The other case revealed retinal neovascularization secondary to proliferative diabetic retinopathy in the eye with choroidal hemangioma.

Key words: Choroidal hemangioma, proliferative diabetic retinopathy, retinal cavernous hemangioma, retinal neovascularization, retinal vascular abnormalities, Sturge–Weber syndrome

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Sturge–Weber syndrome (SWS), also known as encephalo-trigeminal angiomatosis, includes the classic triad of nevus flammeus of the face, ipsilateral leptomeningeal angiomatosis, and ocular involvement.[1] The facial nevus flammeus can involve the eyelids and conjunctiva. Diffuse choroidal hemangioma has been reported in 53–71% of cases which causes choroidal thickening and gives the fundus a diffuse red-orange color.[1] Retinal vascular abnormalities are rare and include vascular tortuosity, arterio-venous malformations.[2,3] We report two cases of unusual retinal vascular anomalies associated with diffuse choroidal hemangioma in SWS.

Case Reports

Case 1

A 21-year-old man was referred to our clinic with a diagnosis of SWS and glaucoma. He had undergone trabeculectomy with retinal cryopexy and laser photocoagulation in the left eye previously. He was seen to have a port wine stain on the left side of the face with hypertrophy of the lip and palate. Visual acuity was 20/20 in the right eye and 20/80 in left eye. Intraocular pressure was 10 mmHg in both the eyes. A thin, cystic bleb was seen superiorly. The right eye fundus examination was unremarkable except for vertical cup disc ratio of 0.6. Diffuse orange red color was noted on left eye fundus examination [Fig. 1]. The cup disc ratio was 0.8. Chorioretinal atrophic patches were seen in the superior and temporal periphery. Dilated tortuous vessels with terminal dark red saccules were witnessed in temporal and inferotemporal quadrants. Fundus fluorescein angiography showed characteristic "fluorescein cap" lesions suggestive of cavernous hemangioma of the retina. On optical coherence tomography subfoveal choroidal thickness was 675 microns in the right eye and 725 microns in the affected left eye. The magnetic resonance imaging (MRI) of the brain was normal. The family history was negative for both the diseases. Since the patient's vision and intraocular pressure were maintained, he was observed further without any intervention.

Case 2

A 51-year-old gentleman, known to have type 2 diabetes mellitus for 15 years, reported to our clinic with decreased vision in the right eye of 10 days duration. He had a port wine stain on the left side of the face associated with hemi-facial hypertrophy [Fig. 2]. The visual acuity was 20/80 in the right eye and 20/20 in the left eye. The right eye showed inferior vitreous hemorrhage due to proliferative diabetic retinopathy (DR). The left eye showed reddish orange fundus color due to a diffuse choroidal hemangioma. Furthermore, the superotemporal and temporal retina revealed the presence of large, flat neovascularization (NVE) [Fig. 3]. The fluorescein angiography revealed extensive capillary non-perfusion in addition to many other NVEs throughout the fundus [Fig. 4]. The intraocular pressure in both the eyes was 18 mmHg. The ultrasonography as well as the swept-source optical coherence tomography confirmed diffuse thickening of the choroid. The subfoveal choroidal thickness was 341 and 467 microns in the right and left eye, respectively [Fig. 4]. A diagnosis of proliferative DR with SWS was made. Systemic examination and MRI of the brain was found to be normal.

Patient was advised panretinal photocoagulation in both eyes. However, after the second sitting of panretinal photocoagulation, the left eye developed peripheral shallow choroidal detachment along with shallow inferior exudative retinal detachment [Fig. 3]. Further laser was deferred and the patient was given topical steroid eye drops. This exudation resolved spontaneously over the next two weeks.

Discussion

Traditionally, SWS includes vascular tumors of the choroid with normal retinal vasculature. There are sporadic reports of retinal vascular anomalies such as dilated retinal veins and retinal arteriovenous communications associated with SWS. Gerwin and Char[3] described a 7-year-old girl with features of SWS who also had dilated retinal vessels leading to an involuted angiomatous lesion in the affected eye. Allen and Parlette[4] reported a patient with port wine stain and a presumed Coats disease in the affected eye. However, in their patient no active telangiectatic or exudative lesions were seen. But it was presumed from a previous history of cryotherapy done elsewhere that the patient might be harboring Coats disease.

Diffuse choroidal hemangioma and cavernous hemangioma of the retina are rarely seen together. Cavernous hemangioma is usually considered to be the only ocular manifestation of this autosomal dominant condition. Sarraf *et al*.[5] described a family with neuro-, oculo- cutaneous phacomatosis referred to as cavernoma multiplex. Members of one family showed cerebral cavernous hemangiomas, cutaneous hemangiomas and choroidal hemangiomas. One member had a retinal cavernous hemangioma alone. The rest of the members had cerebral and cutaneous hemangiomas. It was thought to reflect a variable expression of the disorder and the causative gene was thought to be in the locus of 7q11–q22. However, none of the affected members had both retinal cavernous hemangioma and a choroidal hemangioma together and certainly not in the same eye.

Our patient shows the unique features of a retinal cavernous hemangioma and a diffuse choroidal hemangioma in the same eye. No genetic studies could be done for this patient, but the family history did not reveal any familial diseases such as SWS or related syndromes or retinal cavernous hemangioma. However, the presence of retinal cavernous hemangioma or a choroidal hemangioma should alert one to search for possible systemic and familial involvement.

The second patient in this report showed presence of high risk proliferative DR in the eye affected with diffuse choroidal hemangioma. This association has not been reported in the literature. The retinal pigment epithelium (RPE) overlying the choroidal hemangioma can show either hyperplasia or atrophy. Shields and Shields[6] found thinning and irregularity of RPE in 40% of cases with choroidal hemangioma. These changes accord protection from the development of DR. As a result one rarely sees DR in extreme cases of retinitis pigmentosa, pathologic myopia, etc., In a population based cohort study, Chen *et al*.[7] found no significant association of retinitis pigmentosa with DR. They concluded that the loss of the outer retina might decrease oxygen consumption by the photoreceptors and thus prevent DR.

Arora *et al*.[1] showed a thickened choroid in the affected eyes of patients with SWS which may remain asymptomatic, or develop subretinal hemorrhage and serous retinal detachment

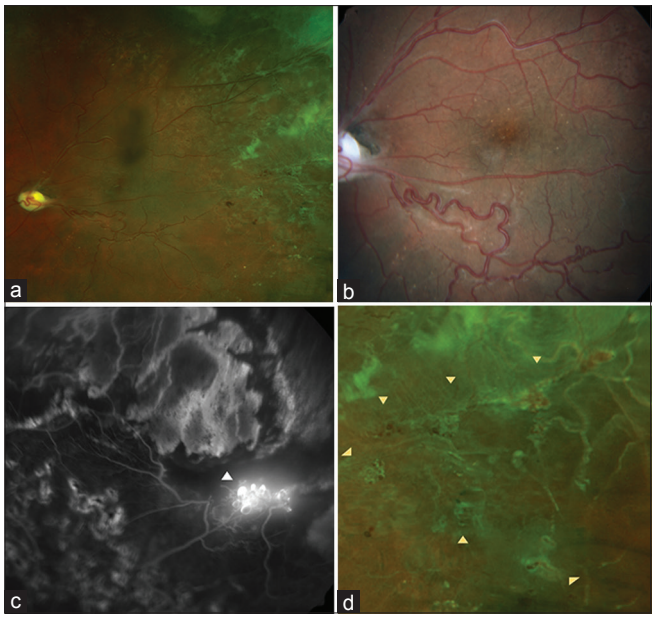

Figure 1: Fundus images of case 1 showing widefield image of the retina (a) with areas of cryo atrophy and laser marks. The posterior pole shows reddish orange appearance due to choroidal hemangioma. Note the dilated and tortuous inferior retinal vessels (b). The temporal area, magnified (c), shows multiple bunches of saccular lesions of the retinal vessels (arrowheads). The corresponding fluorescein angiogram shows the typical fluorescein cap appearance of the lesion (white arrowhead) (d)

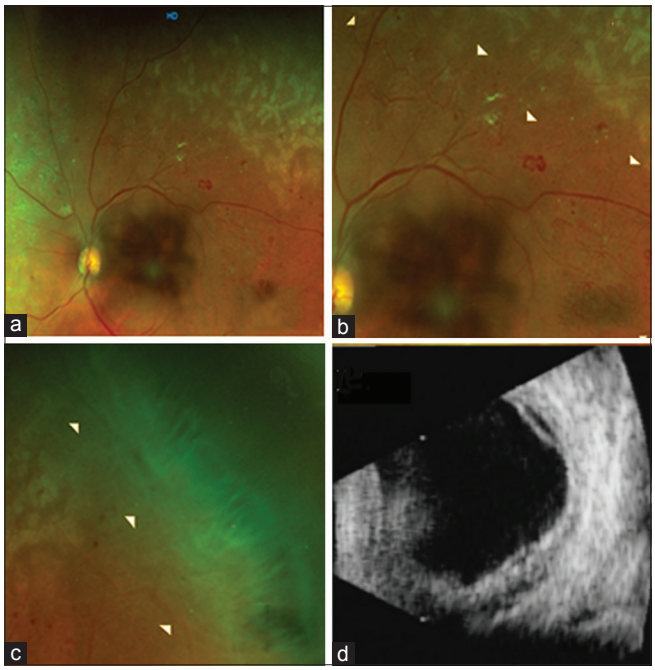

Figure 3: Widefield image of the fundus of case 2, shows laser marks from the panretinal photocoagulation (a). Note the large, flat neovascularization along the superior arcade (b – arrowheads). Central black opacity is due to posterior subcapsular cataract. (c) Shows peripheral shallow choroidal detachment (arrowheads) which is better appreciated on ultrasonography (d)

leading to vision loss.[3] There is a higher risk of developing exudative retinal detachment either spontaneously or induced

Figure 2: External photo of the face, case 2. Note the port wine stain on the left side along with hypertrophy of the lip and face on the left side

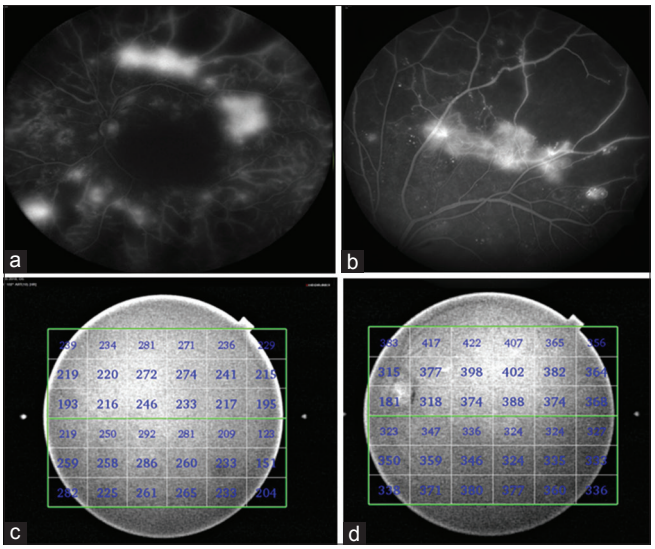

Figure 4: (a and b) Show the fundus fluorescein angiography pictures showing hyperfluorescence due to leakage from multiple areas of retinal neovascularization. Extensive areas of capillary non-perfusion are seen anterior to the neovascularization. (c and d) Show the choroidal thickness map on swept-source optical coherence tomography in the right and left eye, respectively. In the left eye the choroidal hemangioma caused thickening at all points

by drug.[8] Pan retinal laser photocoagulation has been recommended for the management of high risk proliferative DR, however it has been shown to result in increased choroidal thickness and blood flow till after 1 week.[9] Laser photocoagulation in already thickened and vascular choroid may lead to serious retinal detachment. Various modalities of treatment have been suggested including photodynamic therapy, anti-vascular endothelial growth factor antibodies

injections, propranolol, and observation.[10] Our patient resolved spontaneously with observation alone.

Conclusion

In conclusion, we report two cases of SWS with diffuse choroidal hemangioma who also had retinal cavernous hemangioma and retinal NVE due to proliferative DR in the affected eye. Such simultaneous occurrence of retinal vascular abnormalities has not been reported in the literature before.

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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Conflicts of interest

There are no conflicts of interest.

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