

Case presentation and literature review  
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## Introduction

Sturge-Weber syndrome (SWS) or encephalofacial angiomatosis is a rare, sporadic neuro-oculocutaneous disorder, that occurs in approximately one per 50,000 live births. Males and females are equally affected and there is no racial bias.<sup>(1)</sup> Clinical features of SWS may include unilateral facial cutaneous angioma (port-wine stain), buphthalmos, ipsilateral leptomeningeal angiomatosis, epilepsy, focal neurologic deficits (hemianopia, hemiparesis), and a highly variable degree of mental deficiency (with normal intelligence in many affected persons).

In SWS, the most significant, purely vascular anomaly of the eye is a diffuse choroidal hemangioma, in contrast to a well-circumscribed choroidal hemangiomas seen in patients without the syndrome.<sup>(2)</sup>

Although circumscribed choroidal hemangiomas are well documented in cases without SWS, only two previous cases have been reported in patients with SWS, both of which were bilateral in nature.<sup>(4-5)</sup> The following is the third reported case study of a patient with Sturge-Weber syndrome who developed a circumscribed choroidal hemangioma instead of the usual diffuse manifestation.

## Case Photos



**Figure 1.** Case study patient manifesting a facial hemangioma over left face involving the upper and lower lids. This increases the chance of ocular involvement in a patient with SWS. It consists of dilated thin-walled capillaries and venules, most of which are situated in the upper part of the reticular dermis.



**Figure 2.** Left eye shows leukocoria, chemosis and dilated tortuous episcleral vessels. The leukocoria in this case is due to the exudative retinal detachment.



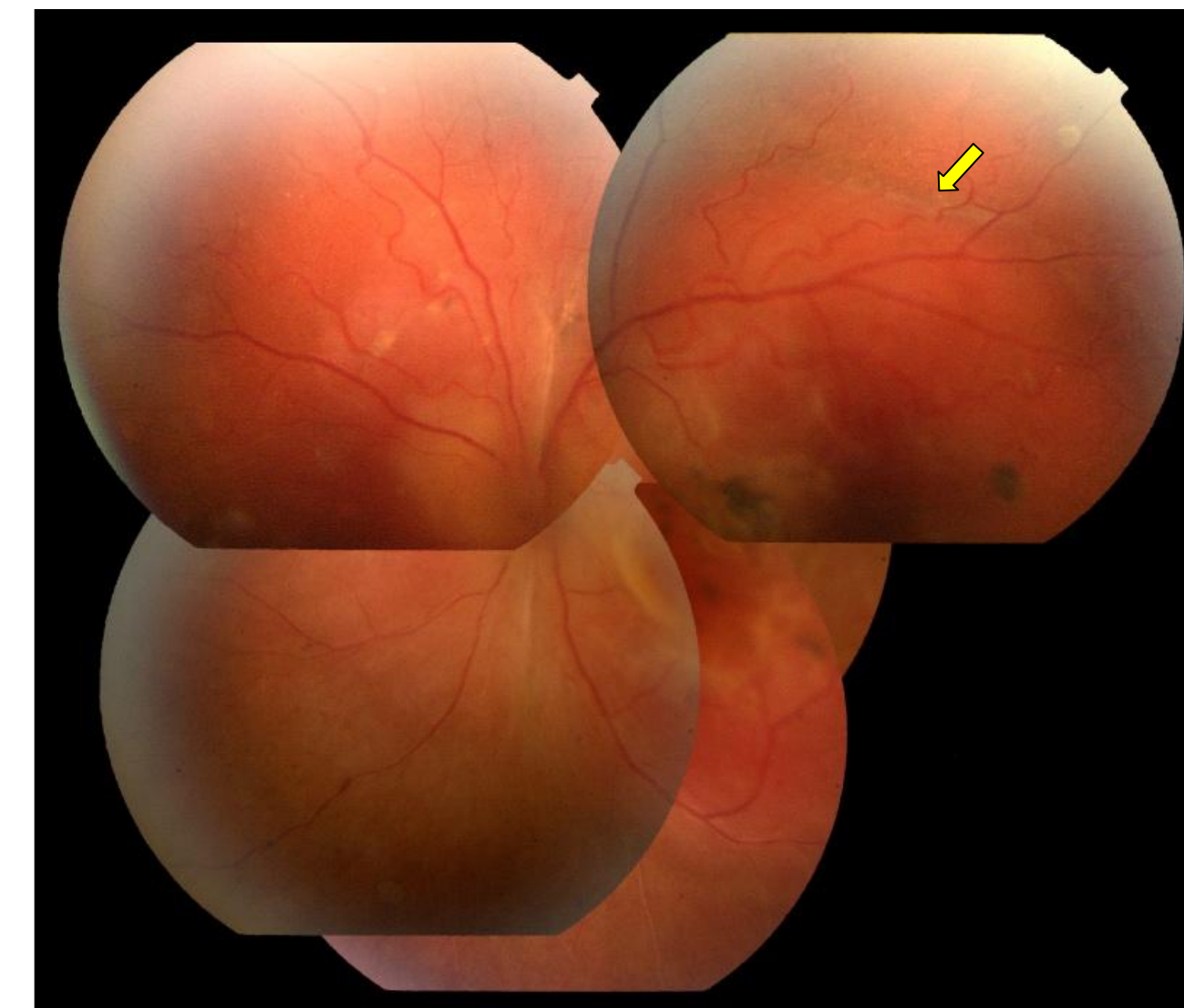
**Figure 3.** Color fundus photo of the patient's right eye with mild elevation of the nasal disc and mild vascular tortuosity.

## Case Presentation

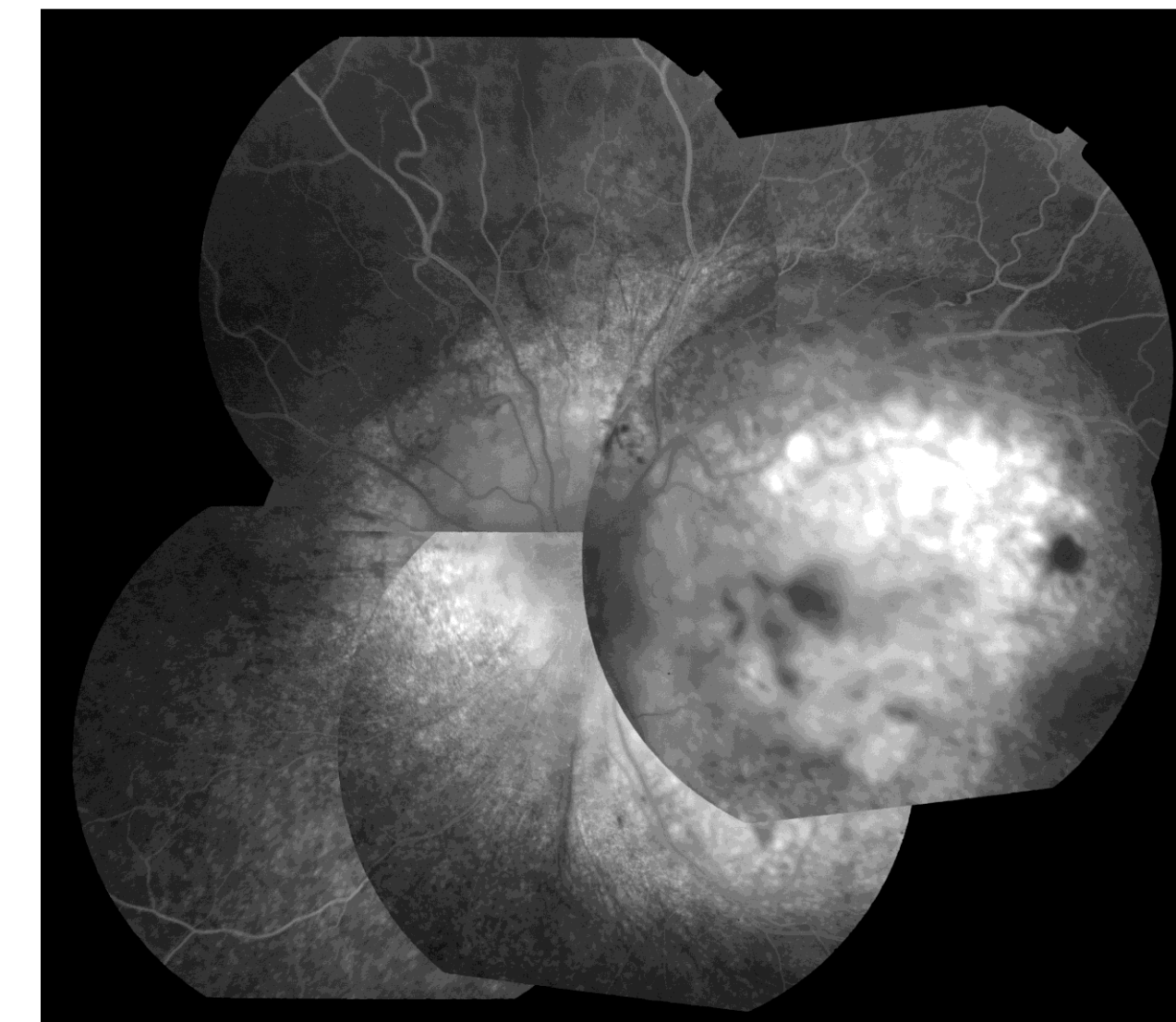
**HPI:** A 19-year-old Caucasian female was referred by her optometrist to the emergency room for evaluation of elevated intraocular pressure (16 OD; 28 OS) and papilledema in her left eye. Approximately one week prior, she began experiencing headaches and sharp shooting pains that built gradually behind both eyes. The vision in her left eye had been poor since childhood but recently was noticing some flashing lights, floating objects and increased loss of peripheral vision. Her left eye carried the diagnosis of glaucoma for which she had previously received treatment but was not currently under treatment or care for this disease. There was no history of eye patching, mass lesions, seizures, or mental retardation. She denied any previous eye injuries or trauma. Past surgical history was positive for a laser procedure OS for unknown reasons. Other personal, family and social histories were noncontributory.

**Exam:** Visual acuity was 20/20 OD and CF at 1 foot OS. She had a 3+ left afferent pupillary defect and decreased confrontational visual fields superior/nasally OS. EOM's were full OU but she had a slight exotropia OS. Intraocular pressures were 23 OD and 31 OS. Slit lamp examination was remarkable for a light red hemangioma of the skin involving the left trigeminal regions of V1 and V2, including the upper and lower eyelids (fig 1). Trace left conjunctival chemosis, dilated episcleral vessels and leukocoria of the left pupil were also evident (fig 2). Cornea was of normal diameter and no clinical evidence of buphthalmos. Anterior chamber was deep with trace flare. Lens was clear and the anterior vitreous face was positive for pigmented cells.

Findings from a dilated fundoscopic examination revealed vitreous without hemorrhage in both eyes. Cup to disc ratio in the right eye was 0.2 with some mild nasal elevation of the optic nerve and vascular tortuosity (fig 3). In the left eye she had fluid surrounding the optic nerve. In the macula we found central pigmentary and degenerative changes of the outer retinal layers as well as an area of elevation that encompassed the entire macula going beyond the arcade vessels, most likely corresponding to an exudative retinal detachment. Beneath this, she had a red elevated area with relatively sharp borders consistent with a choroidal hemangioma. She also had inferior sclerotic vessels in the periphery of the left eye (fig 4).



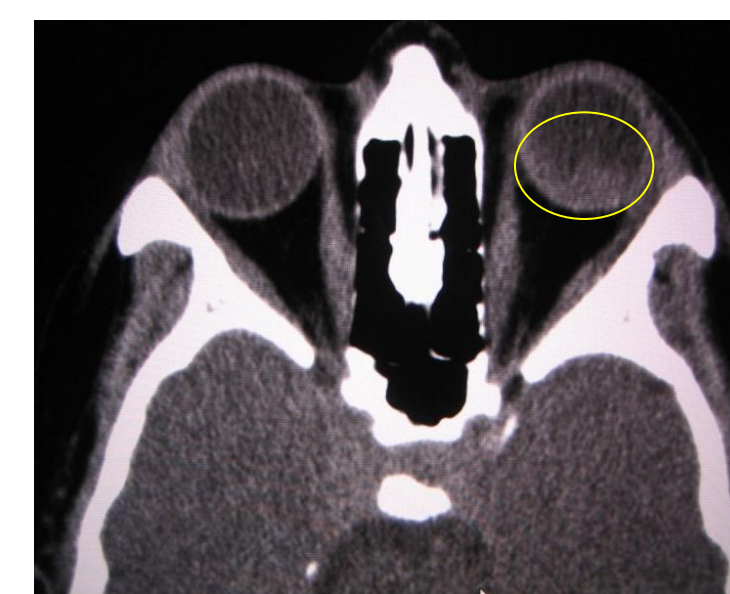
**Figure 4.** Color fundus photo of patient's left eye demonstrating a domed orange/red lesion with pigmentary changes and overlying retinal detachment. Noted is a ring of hyperpigmentation outlining the choroidal hemangioma (arrow).



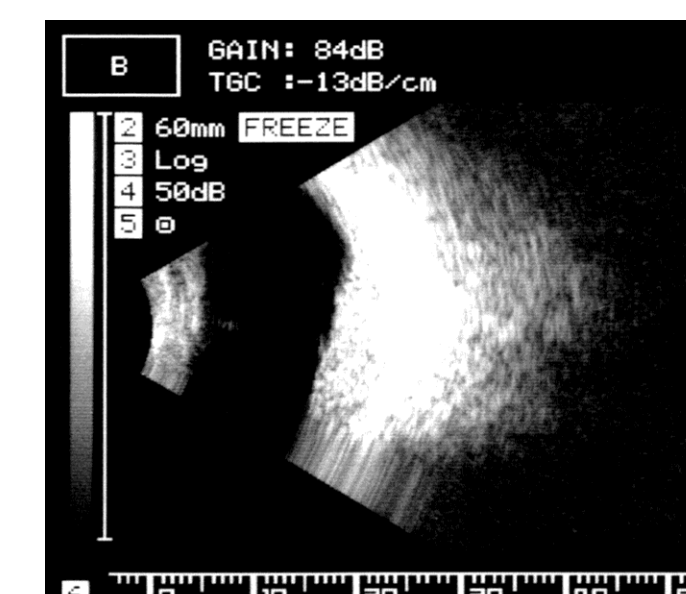
**Figure 5.** Fluorescein angiography of the left eye taken during the laminar phase exhibits choroidal hyperfluorescence of the lesion.

The patient had a CT scan of the brain and orbits which showed a normal cerebrum without atrophy, mass effect or calcification but there was an area of high density posteriorly within the left vitreous (fig 6). All other labs were essentially normal. Echography revealed a left circumscribed choroidal lesion with extensive choroidal thickening on B scan and high internal reflectivity on A scan (fig 7). Fluorescein angiography of the lesion showed dense early central hyperfluorescence and late staining (fig 5). OCT of the lesion illustrated what appears to be both subretinal and sub-RPE fluid in the left eye (fig 8).

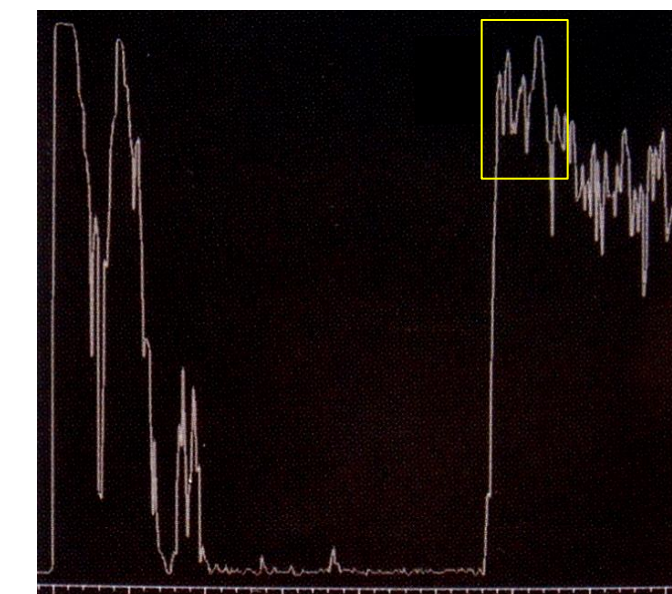
These findings confirmed the diagnosis of circumscribed choroidal hemangioma with an exudative retinal detachment over the posterior pole of the left eye.



**Figure 6.** Hyper-dense area left vitreous (circle) corresponding to an exudative retinal detachment.



**Figure 7.** B scan (left) shows hyperechoic area beneath the retina. A scan (right), shows the high internal reflectivity (box) of the lesion typical of choroidal hemangioma.



**Figure 8.** OCT of patient's left eye noting the retinal detachment and subchoroidal fluid with distortion of the foveal architecture.

## Case Follow-up

Further review of her history and records revealed a previous diagnosis of Sturge-Weber syndrome including its common features of nevus flammeus, choroidal hemangioma and glaucoma. The "birthmark" over the left side of her face had been present since birth. At the age of 10 the eye became red and her vision began to decrease. By age 13, she was diagnosed with glaucoma in the left eye. In 2008, she developed a left choroidal hemangioma for which she received a single treatment of photodynamic therapy (PDT) and was subsequently lost to follow-up until this recent ER visit. Currently, the choroidal hemangioma remains which resulted in an exudative retinal detachment. Her IOP is controlled with aqueous suppressants and she is considering further treatment with PDT. Vision recovery is not expected due to the long history of glaucoma and duration of retinal detachment.

## Discussion

In 1879, William Allen Sturge presented a case of a 6-year-old girl with bilateral facial nevus, congenital glaucoma and vascular malformations of the right eye together with progressive twitching on the left side of her body. In 1922, Parkes Weber was the first to demonstrate the associated intracranial calcifications in this syndrome radiologically. Later, the neurocutaneous syndrome was first discussed as a clinical entity by the ophthalmologist Van der Hoeve.<sup>(5)</sup>

SWS is a vascular disorder with cutaneous, neurologic and ocular manifestations. Most cases with SWS are not life threatening. It is a progressive disease, associated with continuous neurological decline.<sup>(6)</sup> With vigorous control and treatment of symptoms such as seizures, visual problems, paralysis and mental disorder, quality of life can be preserved.

Any portion of the ocular circulation may be affected in SWS with greater occurrence if the skin lesion involves the upper or lower eye lids. Increased conjunctival vascularity gives the eye a pinkish hue. 30% to 70% of patients often develop glaucoma in the affected eye as a result of anterior segment malformation or increased episcleral venous pressure.<sup>(1)</sup> This can sometimes be evident on gonioscopy with blood in schlemm's canal. Onset of glaucoma can be at birth or later in childhood. If IOP is elevated during infancy, buphthalmos can occur.

The retina sometimes shows tortuous vessels and AV communications but the choroid is the site most frequently involved. In SWS, diffuse cavernous hemangiomas predominate. This typically gives the fundus a uniform bright red-orange hue, often compared to tomato catsup. These abnormal choroidal vessels extend to the periphery without underlying choroidal markings. FA shows early hyperfluorescence of the lesion due to rapid filling of its prominent choroidal vasculature followed by late staining. A circumscribed choroidal hemangioma is similar in color but dome shaped and arises in the posterior pole. It is further distinguished by involvement of the RPE and cystic changes of the retinal outer layers. Circumscribed types also have sharply demarcated borders corresponding to compression of the surrounding melanocytes and choroidal lamellae.<sup>(2)</sup> This is seen clinically as a ring of peripheral hyperpigmentation and on FA as blockage of the underlying choroidal fluorescence. Choroidal hemangiomas usually remain silent until adolescence or adulthood, when the patient becomes symptomatic due to a secondary exudative retinal detachment, metamorphopsia, photopsia, micropsia or vision loss.

## Treatment

Treatment of SWS is also three fold; cutaneous, neurologic and ocular. A cutaneous hemangioma may be treated with pulsed dye laser therapy to decrease vascularity and thickening.<sup>(7)</sup> Seizures are of major neurologic concern and are controlled with antiepileptics and partial lobectomy.<sup>(8)</sup> Ocular treatment consists of glaucoma control and treatment of the choroidal hemangioma if symptomatic. Pressure control should begin with aqueous suppressants and if IOP becomes intractable, goniotomy, trabeculectomy or shunt surgery may be required. A choroidal hemangioma may be observed if asymptomatic.<sup>(9)</sup> Several methods have been reported for the treatment of symptomatic choroidal hemangiomas including photocoagulation, cryopexy, radiotherapy, transpupillary thermotherapy, photodynamic therapy (PDT) and anti-VEGF treatment. PDT has shown the most promising effects. Over the past decade several reports have publicized excellent response to PDT, with nearly universal tumor regression and stabilization or improvement of vision among 80% of eyes.<sup>(10)</sup> Recently, reports of anti-VEGF therapy resulting in resolution of exudative retinal detachments caused by choroidal hemangiomas have been published. Vision benefit is limited however by the duration of retinal detachment making it even more important for early diagnosis and treatment.<sup>(11)</sup>

## Conclusion

What makes this case unique is the manifestation of a circumscribed choroidal hemangioma rather than a diffuse type typically found in Sturge-Weber syndrome. This patient displayed all of the classic ocular signs of SWS with the exception of this rare entity. As shown in figures 4 and 5, hyperpigmentation at the periphery of the lesion and the corresponding blockage on FA, delineate its circumscribed nature. This case demonstrates that patients with Sturge-Weber syndrome may also harbor circumscribed choroidal hemangiomas that warrant careful follow-up in order to promptly detect the need for treatment.

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