The Boy With a Growing Shadow in His Vision

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Introduction:

A 14-year-old Caucasian boy presented to our office with a progressive growing shadow in his right eye. We had last seen him when he was 9 years old and had been referred to our office for a chronically red right eye. It had previously been attributed to allergies, and, apart from its appearance, it did not bother him. We diagnosed him with Sturge-Weber syndrome, based on his constellation of physical exam and fundus findings. He had no ocular or systemic complications at the time, so routine follow-up with his general ophthalmologist with monitoring of intraocular pressure was recommended.

Figure 1: Montage of the right eye showing a diffusely dark red choroid from 1 o'clock to 10 o'clock, a reddish orange lesion in the macula, and an inferior retinal detachment.

Now 14 years old, he was having difficulty seeing out of his right eye. In addition to having blurry vision, he reported feeling like a shade was covering the superior aspect of his visual field, and it has been gradually worsening over the past month.

The Exam:

The patient's visual acuity was 20/80 in the right eye and 20/15 in the left eye. There was no afferent pupillary defect. His intraocular pressure by applanation was 15 mm Hg in the right eye and 19 mm Hg in the left eye. His anterior segment examination showed conjunctival hyperemia in the inferior aspect of his right eye. Gonioscopy of that eye showed prominent iris vessels in the inferior angle, without iris or angle neovascularization.

On dilated fundus examination, he was noted to have a well-circumscribed orange-red lesion within the macula and an inferior retinal detachment from 3 o'clock to 9 o'clock (Fig. 1). He also had a diffuse dark red appearance of the choroid from 1 o'clock to 10 o'clock. B-scan ultrasonography of the right eye revealed a diffusely thickened choroid with a focal area of round choroidal elevation in the macula (Fig. 2A). The corresponding A-scan showed medium to high internal reflectivity (Fig. 2B). Fluorescein angiography revealed early stippled hyperfluorescence within the macular lesion with staining in the late phase (Fig. 3). Spectral domain optical coherence tomography (SD-OCT) over the macula showed choroidal elevation with overlying subretinal fluid (Fig. 4A). Interestingly, there appeared to be areas of fluid loculation underneath the retina in a few of the scans (Fig. 4B).

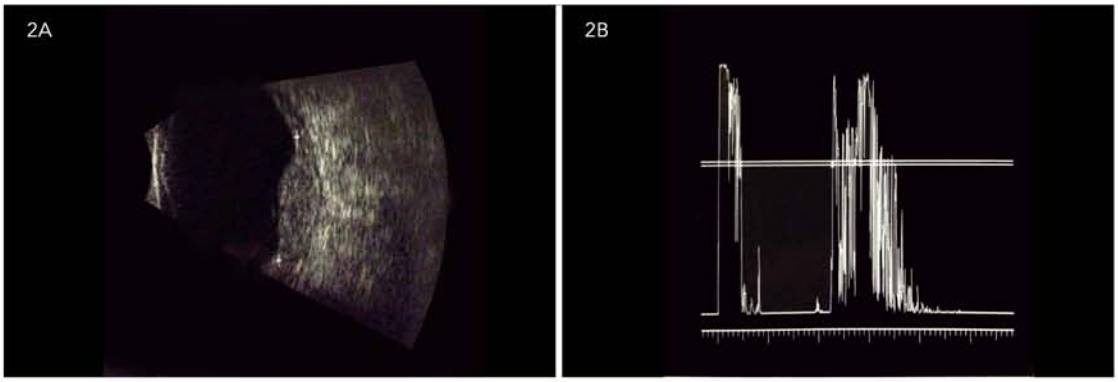

Figure 2A: B-scan ultrasound of the right eye showing a diffusely thickened choroid with focal circumscribed elevation in the macula. Figure 2B: A-scan ultrasound over the lesion showing medium to high internal reflectivity.

On physical examination, the patient had a small nevus flammeus on the right side of his upper lip (Fig. 5), in addition to large patches of pigmented café-au-lait-like lesions on the right side of his chest and back.

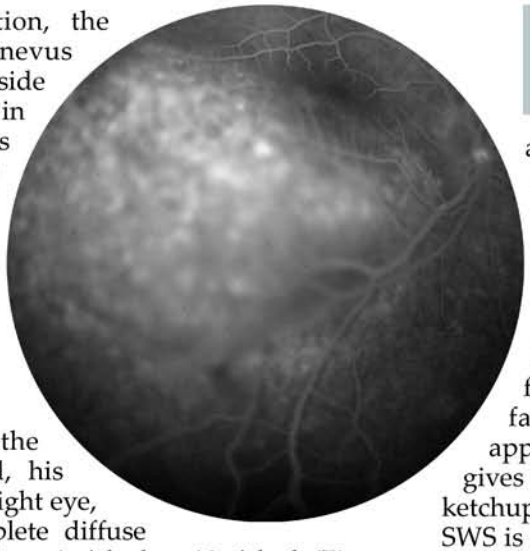

Figure 3: Fluorescein angiogram showing early stippled hyperfluorescence with late staining.

Diagnosis and Treatment:

When we first saw the patient as a 9-year-old, his vision was 20/40 in the right eye, and he had an incomplete diffuse choroidal hemangioma from 1 o'clock to 10 o'clock (Fig. 6). He had normal intraocular pressure, and there was no exudative retinal detachment at that time. We had recommended a screening MRI/MRA to rule out ipsilateral intracranial vascular malformations, but his parents deferred further workup.

Now, 5 years after his initial diagnosis, we believe the patient developed a new focal circumscribed choroidal hemangioma within the macula that caused an inferior exudative retinal detachment. After discussing the treatment options with the patient and his parents, they elected external beam radiation therapy.

He underwent low-dose external beam radiation (≤20 Gy) to his right eye 1 month after being diagnosed with his exudative retinal detachment. Three months after treatment, his vision improved to 20/60 in the right eye. The circumscribed hemangioma in the macula was considerably decreased in size, but there was still a small amount of subretinal fluid remaining inferiorly, outside of the inferior arcade (Fig. 7).

Discussion:

Sturge-Weber syndrome (SWS), also known as encephalofacial angiomatosis, is a congenital neurocutaneous disorder that frequently involves the eye. Unlike other phakomatoses such as neurofibromatosis or tuberous sclerosis, SWS is a sporadic condition with no known inheritance. The incidence of SWS is estimated to be 1 in 20,000 to 50,000 live births.[1]

Possible cause. Recent whole-genome DNA sequencing of visibly affected tissue from patients with SWS revealed a somatic activating mutation in GNAQ1.[2] It is hypothesized that this leads to increased activation signaling, which results in malformed, dysregulated, and progressively dilated blood vessels.

Signs and symptoms. Systemic findings of this syndrome include a cutaneous capillary malformation along the trigeminal nerve (port-wine stain or nevus flammeus), and ipsilateral leptomeningeal vascular malformations. Central nervous system involvement can lead to focal neurological deficits such as seizures, hemiplegia, and mental deficiency. Many patients with SWS, however, have normal intelligence.

In the eye, SWS classically manifests with a diffuse choroidal hemangioma ipsilateral to the facial port-wine stain. The diffuse dark-red appearance of the choroid on ophthalmoscopy gives it the commonly referred to name of a "tomato ketchup" fundus. Although the classic ocular finding in SWS is diffuse choroidal hemangioma ipsilateral to the facial port-wine stain, the most common ocular finding is glaucoma. This can be due to angle development anomalies (usually when patients are younger than 2 years old), or from elevated episcleral venous pressure. In a review of 51 patients with SWS, Sullivan and colleagues found that 71% of patients had glaucoma, 69% had congested conjunctival or episcleral vessels, and 55% had diffuse choroidal hemangioma.[3]

Vision loss in SWS occurs secondary to a hyperopic shift, amblyopia, macular edema, glaucoma, or exudative retinal detachment. Although present at birth, diffuse choroidal hemangiomas often do not cause an

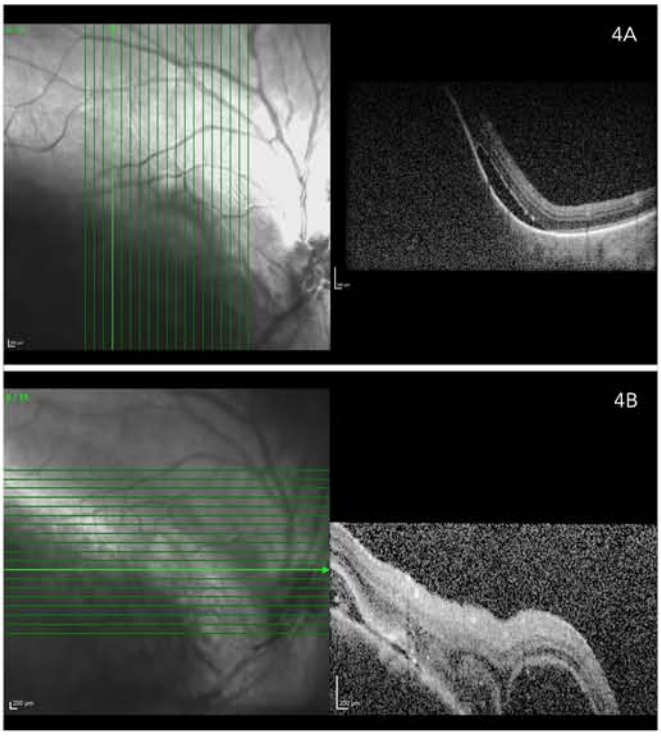

Figure 4A: SD-OCT showing choroidal elevation with overlying subretinal fluid. Figure 4B: Focal areas of loculated subretinal fluid.

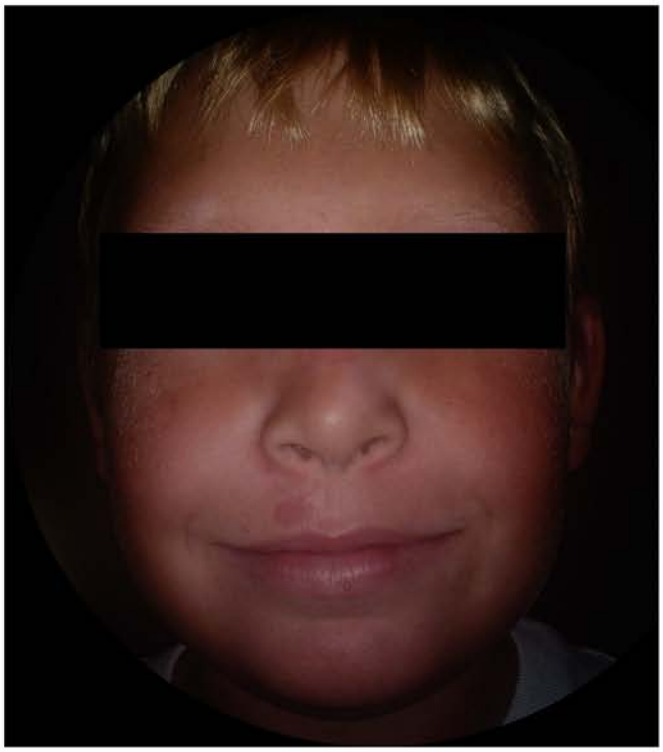

Figure 5: Small nevus flammeus on the right side of the patient's upper lip.

exudative retinal detachment until later in life, with an average age of onset at 8 years.[4] Indeed, patients often won't present with an exudative retinal detachment until adolescence, as the lesion grows with the patient. This was the case in our patient, who had an incomplete diffuse choroidal hemangioma but did not develop an exudative retinal detachment until he reached adolescence.

Treatment. The goal of treatment for diffuse choroidal hemangioma with a concomitant exudative retinal detachment is to cause involution of the lesion and subsequent resolution of subretinal fluid without damaging the overlying retina. Patients without exudative retinal detachment or glaucoma are usually monitored for the development of these complications. Different treatment modalities for these hemangiomas have been reported with varying success, including laser photocoagulation, plaque brachytherapy, photodynamic therapy, and lens-sparing low dose external beam radiation (the latter 2 are the most widely used).[4] Successful involution of diffuse choroidal hemangiomas with oral propranolol or anti-VEGF treatment has also been reported in single case studies,[5-8] but long-term follow-up and larger studies are needed.

Conclusion:

Sturge-Weber syndrome is a rare, spontaneous, congenital neurocutaneous disorder. It commonly presents with vision loss at a young age due to exudative retinal detachment from a diffuse choroidal hemangioma. Prompt recognition and early referral for treatment can help prevent late complications from unrecognized glaucoma or exudative retinal detachment.

We believe our patient presented with a forme fuste, or partial expression, of the syndrome, with an incomplete diffuse choroidal hemangioma, absence of glaucoma, and no mental deficiency. He developed a new circumscribed lesion at adolescence, causing an exudative retinal detachment. The condition responded well to external beam radiation, with lesion regression, resolution of subretinal fluid in the macula, and return of vision to near baseline at 3 months post-treatment. Now at 1 year status post radiation treatment, there are no signs of recurrence, and the patient's visual acuity is 20/30 in the right eye.

Note:

*This case will be featured as the Morning Rounds for the August 2015 of EyeNet.

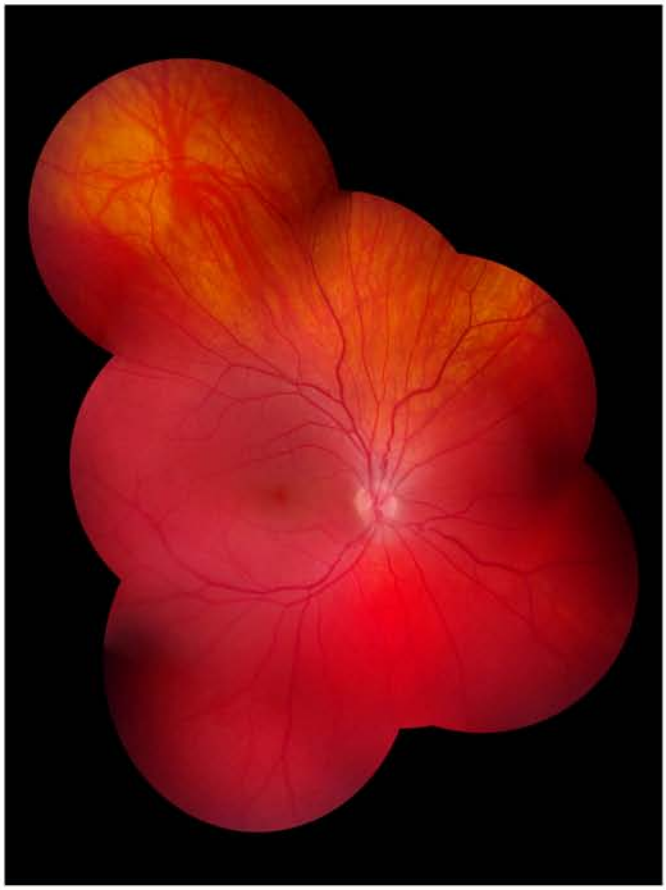

Figure 6: Five years previously, when Jason was 9 years old, we diagnosed him with Sturge-Weber syndrome. At that time, we had noted an incomplete diffuse choroidal hemangioma from 1 o'clock to 10 o'clock.

References:

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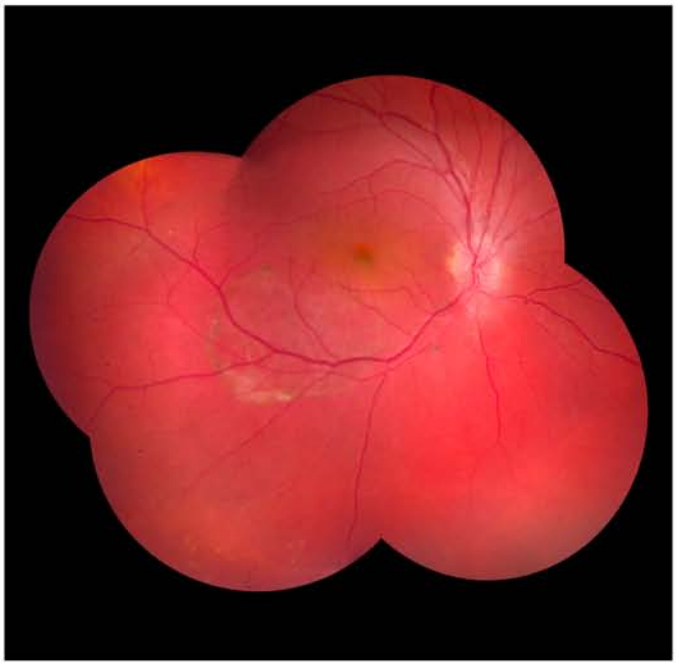

Figure 7: At age 14, he underwent low-dose external radiation for a new macular circumscribed choroidal hemangioma with inferior exudative retinal detachment. Resolution of the subretinal fluid is noted in this fundus montage taken 3 months post-treatment.

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