Chronic Retinal Detachment Secondary to Choroidal Hemangioma in Sturge-Weber Syndrome

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INTRODUCTION

A patient presents for pediatric eye exam complaining of blurred vision in their left eye. Examination reveals port wine stain and reduced visual acuity secondary to chronic serous retinal detachment due to choroidal hemangioma. He is subsequently diagnosed with Sturge-Weber Syndrome (SWS) after further evaluation and diagnostic testing.

CLINICAL FINDINGS

TABLE 1: Entrance testing

Entrance Testing	OD	OS
Visual Acuity	20/20-1 sc	20/100- sc *PH NI **no improvement w/ SRx OS
Pupils	Equal, Round, Reactive, (-)APD	Equal, Round, Reactive, (-)APD
EOMs	Full Range of Motion	Full Range of Motion
Confrontation Visual Fields	Full to Finger Count	Full to Finger Count

TABLE 2: Slit lamp

Anterior Segment	OD	OS
Adnexa	adnexa normal	Port-Wine Stain @ V1 + V2 Dist.
Lids/Lashes	lids and lashes normal	hyperpigmentation @ lateral canthus
Conjunctiva	white and quiet	dilated, tortuous vessels temporal limbus
Sclera	white and quiet	white and quiet
Cornea	normal endothelium, epithelium, stroma and tear film	normal endothelium, epithelium, stroma and tear film
Angles	3-4+ N/T ; open 360 via Gonio	3-4+ N/T ; open 360 via Gonio
Anterior Chamber	deep and quiet	deep and quiet
Iris	normal	normal ; (-) heterochromia
Lens	clear lens capsule, cortex, and nucleus	clear lens capsule, cortex, and nucleus

TABLE 3: Posterior Segment

Posterior Segment	OD	OS
Vitreous	Vitreous clear	Vitreous Clear
Optic Nerve	Flat, sharp, good color	Flat, sharp, elevated area temporal to disc c/w choroidal hemangioma
CD Ratio	0.35/0.35	0.55/0.55
Macula	Flat, no hemorrhages, exudates, pigmentary changes, or macular edema	Pigmentary changes with sub-retinal fluid ; central macular scarring
Vessels	Normal vessels	Normal vessels
Periphery	Flat x 360 degrees, no RD, no holes	Flat x 360 degrees, no RD, no holes
IOP	17 mmHg	17 mmHg

TABLE 4: Additional Testing

Additional Testing	OD	OS
Gonioscopy	Open to CB 360 ; (-) blood / angle abnormality	Open to CB 360 ; (-) blood / angle abnormality
OCT	Healthy RNFL ; (-) RD ; See Figure 3.	See Figure 3.
B-Scan	Normal	See Figure 5.
Fluorescein Angiography / OCT-A	Normal	See Figure 4. Grossly distended choroidal macro-vessels on OCT-A

Computerized Tomography	No intracranial calcification noted. However, hyperreflective in posterior left globe.

FIGURE 1
Anterior Segment Photos
External Photos. A.) Shows Port-Wine Stain (PWS) affecting the left side of the patient's face. B.) PWS affecting the V-1 and V-2 distribution of the Trigeminal Nerve. In addition, hypertrophy of lesion can be noted.

FIGURE 2
Posterior Segment Photos
Posterior Segment Photos : A.) OD reveals healthy optic nerve and macula with no SWS involvement. B.) OS shows healthy optic nerve with circumpapillary diffuse choroidal hemangioma with fibrosis + pigmentary changes involving the macula.

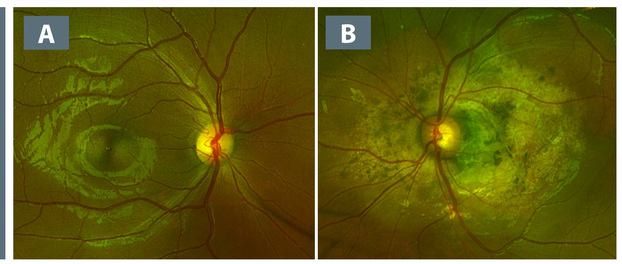


FIGURE 3
OCT – A.) Macular scan shows chronic serous sub-macular retinal detachment secondary to diffuse choroidal hemangioma B.) ONH scan reveals OD within normal limits, while OS shows deep cup with no significant RNFL thinning. Significant artifact limiting scan reliability OS due to elevation caused by hemangioma.

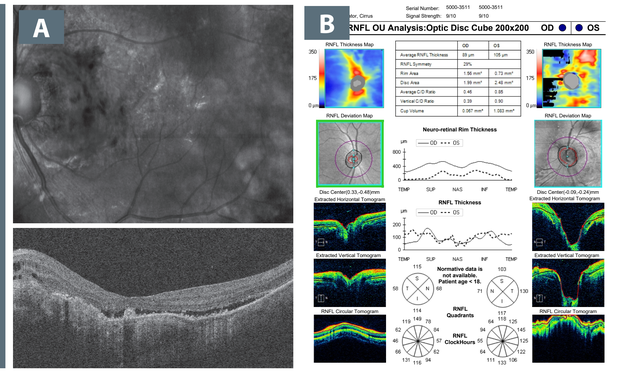


FIGURE 4
Fluorescein Angiography
Fluorescein Angiography OS only, A.) Early extensive peripapillary and sub-macular leakage from abnormal choroidal vasculature. B.) Late peripapillary staining of circumpapillary and sub-macular fibrosis

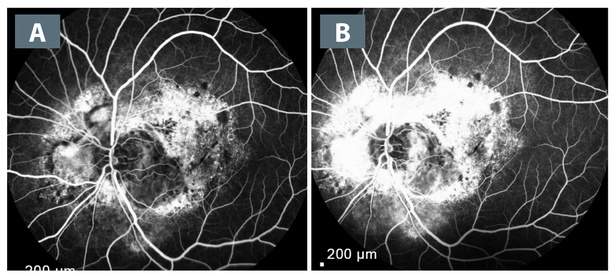
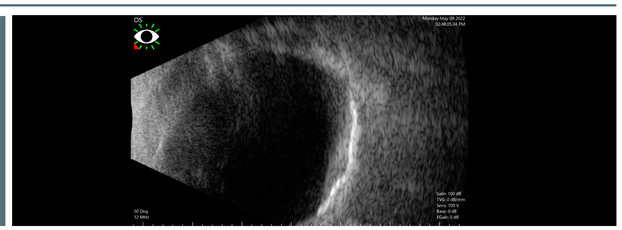


FIGURE 5
B-Scan OS only shows hyperreflective lesion (particularly inferiorly) surrounding optic nerve with increased choroidal thickening.



DIAGNOSIS & DISCUSSION

SWS Type 2: facial port-wine stain (PWS) with no leptomeningeal involvement without glaucoma

SWS is a rare, sporadic neuro-oculo-cutaneous syndrome characterized by leptomeningeal angiomatosis, facial PWS, neurologic problems, and ocular complications. The extent of PWS, leptomeningeal, and ocular involvement depend on the moment in which mutation of the GNAQ gene occurs. This is thought to cause impaired development of neural crest cells. Ocular complications include pathological changes involving the lid, cornea, anterior chamber, choroid, and retina. Due to our patients' age and lack of seizure history, it was presumed neurologic sequela were unlikely. 75-90% of patients with SWS develop seizures before the age of 2. If patients are diagnosed with SWS after 2, the need for MRI is debatable in asymptomatic patients, since late-onset seizures are much less likely to occur in early adolescence. However, management of serous RD secondary to choroidal hemangioma and close monitoring for late-onset glaucoma are critical. 30-70% of patients with SWS develop glaucoma and 40-50% have choroidal hemangiomas. Subsequent glaucoma can be difficult to manage with medical therapy, and often requires surgical intervention.

Glaucoma pathogenesis is complex in SWS patients due to interplay of various interlinked mechanisms that change with aging. Congenital or early-onset glaucoma typically angle dysgenesis plays a pivotal role, while in late-onset glaucoma an increase in episcleral venous pressure is the driving force.

TREATMENT

- Recommended retinal consult to determine the need for PDT treatment of choroidal hemangioma. Due to the location of hemangioma, the retinal specialist recommended annual monitoring.
- No topical aqueous suppressants were recommended at this time due to normotensive IOP, healthy optic nerves, and stable RNFL/GCA on repeat OCT. Close monitoring every 4 months with IOP check and annual OCT-RNFL + visual field recommended.
- Discussed possibility of pulsed dye laser (PDL) treatment for PWS. Family will pursue dermatology referral through their PCP.
- Emphasized importance of monitoring for seizure episodes, migraine-like headaches, and episodes like cerebrovascular events.

CONCLUSION

SWS is a rare syndrome that can have a severe impact on visual acuity. It is critical for the optometrist to be aware of all the ocular and neurological complications that can be associated with PWS in a pediatric patient. Co-management with neurology and ophthalmology are critical due to the significant systemic complications associated with SWS.

REFERENCES

Available upon request.

CONTACT

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