

## Variant of Sturge-Weber Syndrome

*Annals of Ophthalmology*

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### ABSTRACT

A case of Sturge-Weber syndrome in a 27-year-old man involved only the skin and left eye, without any features suggestive of central nervous system involvement. Additionally, this patient had ciliary body angioma and orbital angioma in the affected eye, a finding not known to have been previously reported in the literature.

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Phakomatoses are a group of hereditary disorders characterized by formation of hamartomas in the eye, skin, nervous system, and viscera. The disorder occurs due to interference in migration and differentiation of embryonic cells resulting in proliferation.<sup>1</sup> Various disorders described under this entity are neurofibromatosis, angiomas of retinae, encephalotrigeminal angiomas, tuberous sclerosis, oculodermal melanosis, basal nevus syndrome, and ataxia telangiectasia. Glaucoma is a common occurrence in phakomatoses. Hence, its early detection and treatment is of utmost importance.<sup>2</sup>

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We herein report a case of Sturge-Weber syndrome having involvement only of the skin and 1 eye, without any features suggestive of central nervous system involvement. Additionally, this patient had ciliary body angioma and orbital angioma in the affected eye, which has not been previously reported in the literature, to the best of our knowledge.<sup>3</sup>

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### Case Report

We herein describe a 27-year-old man, who presented with painless decrease of vision in his left eye that gradually progressed for 3 years. On examination, the patient had a left-sided port-wine stain involving the forehead, upper eyelid, and part of the nose (Fig 1), which he reported had been present since birth. Visual acuity was 6/6 in the right eye and counting fingers in the left eye. There was fullness in both eyelids, with 2-mm ptosis on the affected side. The eyeball was protruded on the left side by 2 mm (Fig 2), with 10 prism diopters of exotropia. Conjunctival and episcleral vessels were dilated, tortuous, and engorged all around the limbus. The cornea and anterior chamber appeared normal, but the iris was hyperchromic. There was an afferent pupillary defect in the left eye. Gonioscopy revealed an open angle with a localized area of indentation at the 2-o'clock position. On fundus examination, the cup-to-disc ratio was 0.9:1 with deep cupping and bayoneting of retinal vessels, with positive laminar dot sign. There was no evidence of choroidal hemangioma on indirect ophthalmoscopy and fundus fluorescein angiography. Intraocular pressure was 30 mm Hg in the left eye. Visual field examination was not possible in the left eye due to poor vision. Visual field findings in the right eye were normal.

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Ultrasonography of the left eye detected a soft-tissue-density mass measuring 8 × 6 × 6.5 mm in the ciliary body region at the 2-o'clock position, which showed a small calcification posteriorly (Fig 3). Color Doppler ultrasonography revealed increased vascularity of the lesion (Fig 4). These findings were suggestive of a ciliary body hemangioma. Color Doppler ultrasonography showed a hemangioma in the left orbit just lateral to the medial rectus muscle, with increased venous flow (Fig 5). The computed tomographic (CT) scan of the orbit showed a 12 × 6-mm

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~~lesion of~~ irregular and variegated density in the left orbit between the medial rectus ~~muscle~~ and optic ~~nerve; the~~ contrast enhancement ~~was~~ suggestive of orbital hemangioma (Fig 2). The CT scan of ~~the~~ brain was essentially normal. The chest ~~x-ray; the ultrasonograms of the abdominal organs; and results of the ear, nose, and throat~~ examination did not show any evidence of hemangioma.

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Based on the above findings, the patient was ~~thought to have~~ a rare case of Sturge-Weber syndrome with no involvement of ~~the~~ central nervous system despite involvement of the upper eyelid. Intraocular pressure was controlled with topical timolol ~~maleate, 0.5% twice a day, and oral acetazolamide, 250 mg 3 times a day.~~ Subsequently, oral acetazolamide was replaced by dipivefrin hydrochloride ~~eyedrops, 0.5% twice daily~~ for 2 weeks. Four months after follow-up, the intraocular pressure was normal.

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