Original Research Paper Medical Science

Sturge Weber Syndrome Type 1- A Rare Case Report

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ABSTRACT

Sturge–Weber angiomatosis is a rare, nonhereditary developmental condition characterized by a hamartomatous vascular proliferation involving the tissues of brain and face. A report of a case with facial port wine stains, gingival overgrowth, and dilated ocular vessels is described.we are reporting a case of young girl of sturge weber syndrome type 1 with glaucoma her right eye, that was managed successfully with surgery

KEYWORDS | Sturge Weber Syndrome,Glaucoma,Tram Track Calcification

Introduction-

Sturge Weber Syndrome occurs in 1:50,000 live births.1Nearly one third of patients with SWS will have glaucoma in some part of their life .2 There are three variants of SWS.3 Type I - is classical type in which there is portwine stain, leptmeningeal involvement and glaucoma. Type 2- Glaucoma with leptmenigeal involvemernt. Type 3- Only leptomeningeal involvement. We report a case of 12 years old female patient who presented to us with symptoms of watering and photophobia from right eye and later was diagnosed as a case of Sturge Weber Syndrome Type I.

Case Report -

12 year old female patient presented to us with history of watering and photophobia from right eye since 2 months. Past history revealed she was born at full term normal delivery with portwine stain on the the right side of face. At the age of 3 months she had developed seizure which was controlled medically. She again developed seizure at the age of 15 months. At this time CT cranium was done which showed small calcification on the right occipital region. She was put on antiepileptic drugs .Ophthalmic investigations were at that time were within normal limits. Her parents also say that she had disinterest in reading. There were no complaints from left eye. On general examination there was large portwine stain on the right side of face involving the right eye upper and lower eye lid with extending to left side just below the left eye(figure-1).

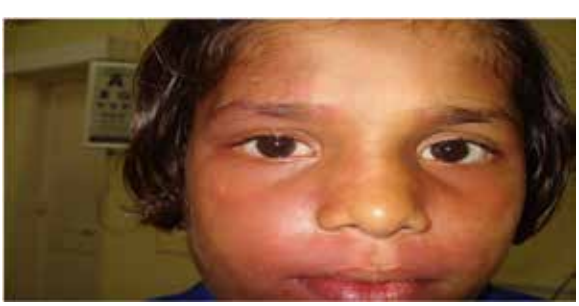

Figure 1-clinical photograph of patient

Her milestone is normal for her age. Ophthalmic examination shows both eye pupil round and reacting. Visual acuity right eye 6/24 corrected to 6/9 and left eye 6/9.. Both eye conjunctiva, cornea, anterior chamber and lens were within normal limits. Fundus examination right eye shows C: D ratio .8 with thinning of neuroretinal rim with nasalization of vessels.(Figure 2)

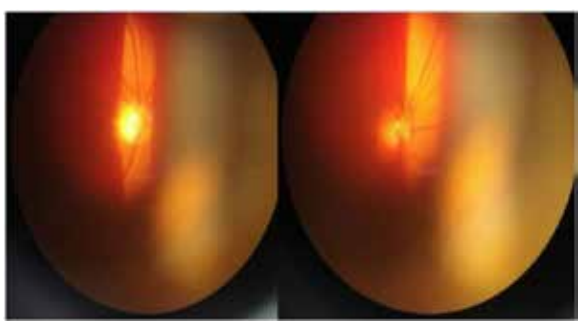

Figure 2-Right eye showing large C D ratio.

Macula within normal limits. Left eye fundus shows C: D ratio .4 with healthy neuroretinal rim. Macula within normal limits. Intraocular pressure right eye was 46 mm of Hg and left eye 18 mm of Hg. Goniscopy both eye shows open angle grade III in all quadrants. CTscan cranium shows tram tack calcification in the right postero-parieto-occipital region with focal atrophy (figure -3).

Figure 3 -CT SHOWING CLASSICAL TRAMTRACK CALCIFICATION

The patient was initially started on topical beta blockers which show no response in 4 weeks. Her IOP remain 44 mm of Hg. Then she tried with combination of prostaglandins analogue and topical beta-blocker with minimal drop in IOP. After that trabeculectomy along with MMC was performed in her right eye . The pressure is reduced to 26 mm of Hg on the first day. She was again started on topical beta blockers (Timolol 0.5% eye drops) .After one month her intraocular pressure were 16 mm of Hg .In one year follow up ,she is maintaining IOP under 18 mm of Hg

Discusssion-

Sturge-Weber Syndrome,sometimes referred to as encephalotrigeminal angiomatosis,occurs ophthalmic and maxillary distribution of trigeminal nerve. It is rare congenital neurological and skin disorder. It is one of the form of phakomatosis and is often associated with portwine stain of face,glaucoma,seizure,mentalretardation,and ipsilateral leptomeningeal angioma. Glaucoma is most often congenital and ipsilateral to the facial nevus, especially if it involves the upper eyelid or conjunctiva.4 Glaucoma in SWS is produced by mechanical obstruction of the angle of the eye, elevated episcleral venous pressure, or hypersecretion of fluid by either the choroidal hemangioma or ciliary body. The anterior chamber angle abnormality is consistently seen in the infantile glaucoma cases in SWS, while increased episcleral venous pressure may have a key role in late-onset glaucoma cases in SWS. Decreased vision and blindness result from untreated glaucoma, with increased IOP leading to optic nerve damage. An acceptable range of IOP is 10-22 mm Hg.Glaucoma in these cases is difficult to treat because these are resistant to antiglaucoma drugs and surgery has high complication rate as before performing any surgery ,IOP has to be in normal range. Complication can occur in the form of expulsive hemorrhage,choriodal detachment. Sullivan et al reviewed ocular abnormalities in 51 patients with SWS .5 Of these, 36 (71%) had glaucoma, with onset before age 24 months in 26 patients; 35 (69%) had conjunctival or episcleral hemangiomas; and 28 had choroidal hemangiomas. With time, choroidal hemangioma may cause other secondary changes such as retinal pigment epithelium degeneration, fibrous metaplasia, cystic retinal degeneration, and retinal detachment. Also retinal vascular tortuosity, iris heterochromia, optic disc coloboma, and cataracts have been seen in patients with SWS. Another feature is Neurologic and developmental morbidity. This includes seizures, weakness, strokes, headaches, hemianopsia, mental retardation, and developmental abnormalities. The development of seizures and the age of onset may correlate with the degree of neurologic involvement. Neurologic dysfunction increases with bilateral PWS. Patients may experience complications related to refractory seizures and anticonvulsants, visual loss and blindness from glaucoma, cosmetic deformities, and other manifestations of soft-tissue involvement.

Conclusion-

We have reported a case of Sturge Weber Syndrome Type 1 with Glaucoma which was refractory to medical treatment but managed successfully with surgery. Surgery in these cases is difficult because of thin sclera and chances of choroidal Hemorrhage.

Conflicts of interest – Author have nothing to declare.

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