

Sturge-Weber syndrome with involvement of chest dermatomes and unilateral buphthalmos: A rare association

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Abstract. Sturge-Weber syndrome is a rare neuro-cutaneous disease presenting as port- wine stain over distribution of trigeminal nerve. There is a wide variability in manifestations of the disease. Associated malformations in the form of buphthalmos is a rare occurrence with this disorder. We report a 2 months old infant who had bilateral port- wine stain over face and extending over the upper part of chest. There were no seizures and developmental milestones were normal. The patient had associated buphthalmos in the right eye. The intraocular pressure in the right eye was raised while it was normal in left eye. Cranial CT scan did not reveal any calcifications. In conclusion, Sturge- Weber syndrome may not present clinically in its typical form. The presence of buphthalmos is to be looked especially when there is extensive bilateral port-wine stains.

Key words: Bilateral port-wine stains, buphthalmos

1. Introduction

Sturge-Weber syndrome (SWS) is a rare oculo-cutaneous disorder, manifesting as facial capillary haemangioma and lepto-meningeal abnormalities which in turn leads to central nervous system (CNS) affiliations like seizures, behavioural and developmental disorders. The CNS involvement is considered as a hallmark of the disease (1). Its exact incidence is not known. It has no racial or gender predilection. Angiomas of SWS is due to failure of regression of a vascular plexus around the cephalic portion of neural tube resulting in residual vascular tissue which forms angiomas of leptomeninges, face and ipsilateral eye (2). Buphthalmos typically occurs when port -wine stains involve eyelids. It can develop at any age, is usually unilateral and

ipsilateral to port wine stain (3). But bilateral presentations associated with chest dermatomes and unilateral buphthalmos is rare entity.

2. Case report

A two months old male child, presented with bilateral port-wine stains and over right upper chest area to a tertiary care hospital in north India. The infant was born as a normal, full term, uneventful and home delivery in a rural region. The right eye of the patient had central corneal opacity (Fig. 1) which according to the patient's mother had increased in size after birth. The patient was examined under sedation and the right eye had a horizontal and vertical corneal diameter of 14 mm and 13 mm, respectively. A hand held slit lamp showed the opacity to be stroma deep and presence of haab's stria. The anterior chamber was deep and the iris and pupil appeared normal.

Fundus details were not visible because of the opacity. The left eye had a horizontal corneal diameter of 12mm and vertical corneal diameter of 11mm. The cornea was clear, anterior chamber was deep and the iris and pupil had no abnormalities. The optic nerve head seen by a

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Fig. 1. Photograph of face and trunk showing bilateral port-wine stains over face and right upper chest with right eye corneal haziness.

Direct ophthalmoscope appeared normal. The Intraocular pressures (IOP) measured by a hand held Perkins tonometer were 22mm of mercury and 16mm of mercury in the right and left eye, respectively. The port-wine stain was present in the distribution area of the ophthalmic and maxillary division of the trigeminal nerve on both the sides of the face. It was also present on the chest dermatomes on the right side (Figure 1). The CT-scan of head did not reveal any abnormality. The child was followed up to one year of age and all the milestones were normally attained. A diagnosis of Sturge-Weber syndrome with distribution over chest dermatomes and buphthalmos was made. The IOP was lowered using topical beta blocker initially for a week, and later on trabeculectomy with Mitomycin-C was done in the right eye. The left eye was normal till last follow up. Informed consent was taken from parents of the child.

3. Discussion

Sturge-Weber syndrome is a neurocutaneous disorder with variable presentations having facial and lepto-meningeal angiomas. It has no definite genetic predisposition. The incidence is 1 in

50,000 live births (4,5). Buphthalmos has been reported in these patients with CNS involvement (6). However, the present case had bilateral port-wine stains and no CNS involvement. The extra facial locations of port-wine stains such as on chest or torso can also occur (7). Seizures with onset below one year of age are more common with bilateral port-wine stains. However, seizures were absent in our case. This indicates that despite presence of large angiomas, the 'vascular steal phenomenon' has not been able to show its effect (8). The mile stones were normal; further suggesting that there has been no CNS involvement.

The presence of corneal haziness with raised IOP indicates that glaucoma can be associated and it is because of maldevelopment of anterior chamber angle structures and increased secretion of aqueous humor by choroidal hemangioma, rather than due to raised episcleral venous pressure. This view is also supported by the fact that glaucoma was unilateral despite facial angiomas being present on both sides of the face. The early detection and treatment of glaucoma is of utmost importance in order to save the vision of patient.

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