Sturge Weber Syndrome – Case report

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Abstract:

Sturge weber syndrome is a rare sporadic condition of mesodermal phacomatosis, also called encephalotrigeminal angiomatosis (synonyms : fourth phacomatosis or mother spot), is a neurocutaneous disorder with angiomas that involve the leptomeninges (leptomeningeal angiomas) and the skin of the face (purple colored flat cutaneous haemangiomas), typically in the ophthalmic (V1) and maxillary (V2) distributions of the trigeminal nerve. The hallmark of sturge weber syndrome is a facial cutaneous venous dilation, also referred to as a nevus flammeus or port wine stain (PWS). Because of the rarity, we report here a one & half year old male child who presented with features of the Sturge Weber Syndrome on both side of face.

Key words : Nevus flammeus, Haemangioma, Phakomatosis, Angioma, Sporadic.

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Introduction :

Sturge-Weber Syndrome (SWS) is a mesodermal phacomatosis characterized by a port wine vascular nevus on the upper part of the face, leptomeningeal angiomatosis that involves one or both hemispheres, choroidal vascular lesions associated with glaucoma, early onset scizure, neurologic detoriation and eventual neuro-developmental delay. The syndrome occur almost always sporadically & have no definite hereditary influence. patient with unilateral facial portwine stains of upper eye lid increases the risk of glaucoma and regularly had seizures & hemiparesis of the contralateral side & that is the result of an intracranial haemangioma. Sturge Weber Syndrome appear as a congenital lesion usually benign tumours of blood vessels & have no chance of malignant transformation.

Case Report :

A one & half yrs. old male child, from Komolpur, Comilla with the complaints of photophobia & reddish discoloration on both side of face (upper part), born at 39 weeks of gestation by C-S at local hospital with uneventful antenatal & postnatal period. Just after birth, they saw that Emon had bilateral reddish discoloration on his upper part of face. At the age of 18 days Emon was taken to a local hospital. From there he was referred to NIOH at Dhaka. His parents noticed that their baby can not tolerate the bright light & eyes became enlarge gradually.

His parents has no consanguinity. Developmental history suggested delayed milestones. Physical examination revealed that there was extensive port wine staining over both sides of his face, extending to both lower eyelids & cheeks, enlarged corneal diameter & patient was photophobic. After examination the patient was diagnosed as Sturge weber syndrome & was admitted at BSMMU eye word.

The haematologic, biochemical and urinary laboratory tests were normal. EEG shows multiple epileptiform foci on both sides & voltage suppression on the left hemisphere. Contrast MRI revealed evidence of leptomeningeal angiomatosis in the left fronto parieto temporal region & enlarged ipsilateral choroid plexus.

He was examined under anaesthesia (EUA) & measure corneal diameter (in both meridian was 14 mm), intraocular pressure (IOP 24 mm Hg on R/E & 28 mm Hg on L/E

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) on ophthalmoscopy glaucomatous cupping of the optic disc with tortuous retinal blood vessels more on left side compared to the right. Before surgery he was treated by anti-glaucoma medication (Timolol E/D). Ttrabeculectomy was done under general anaestheia. 1st done in L/E & after one wk. in R/E/ Now he is better than previous & his corneal diameter is decreasing gradually & IOP is normal (10 mm in B/E)& photophobia decreasing.

Fig.– 1

Fig.– 2

Discussion :

Sturge weber syndrome (encephlotrigeminal angiomatosis) is a congenital sporadic phacomatosis. SWS is a rare neurocutaneous syndrome with a frequency of approximately 1 per 50,000. The exact pathogenesis of these syndrome are still not clearly elucidated. There are suggestions that the majority of cases result from somatic mutations involving genes that play significant roles in embryonic vasculogenesis and angiogenesis.

Types :

- Trisystem disease involve the face, leptomeninges and eyes (type-I).
- Bisystem disease involves the face & eyes or the face & leptomeninges (type-II).
- Isolated leptomeningeal angioma (type-III)

Ocular features are -

- Ipsilateral glaucoma (Facial haemangioma of upper eyelid increases the risk of glaucoma).
- Episcleral haemongioma.
- Iris heterochromia, blood in schlemm's canal (seen on gonioscopy)
- Diffuse choroidal haemongioma (produces a uniform red fundus back ground).
- Secondary serous retinal detachment, secondary retinal pigment epithelial alterations (retinitis pigmentosa like picture)

Other features are –

- Port wine stain extending over the area correspond ing to the distribution of 1st & 2nd division of the trigeminal nerve.
- Ipsilateral parietal or occipital leptomeningeal haemangioma may cause contralateral focal or generalized seizures, hemiparesis or hemianopia.
- Subnormal intelligence or mental retardation.
- Cerebral calcifications.

Pathogenesis of glaucoma :

Glaucoma develops in about 30% of patients ipsilateral to the facial haemangioma, especially if the lesion affects the upper eyelid. Elevation of IOP occurs within the first 2 years of life in 60% of glaucoma patients and may result in buphthalmos. In the remainder, glaucoma may develop any time from infancy to adulthood (pathogenesis is controversial and often obscure). In infants, isolated trabeculodysgenesis may be the cause.

Raised episcleral venous pressure (associated with an arteriovenous communication in an episcleral haemangioma) may be responsible in older patients.

a) Medical treatment with topical prostaglandin analogues may be successful.
b) Goniotomy – may be successful in eye with angle anomalies.
c) Combined trabeculectomy with trabeculotomy gives good result in early onset cases.

The rationale is that trabeculotomy addresses the barrier to aqueous outflow posed by a congenital angle anomaly, while trabeculectomy passes the episcleral veins

Diffuse choroidal haemangioma :

It occurs almost exclusively in patients with sturge weber syndrome ipsilateral to naevus flammeus (port wine stain). It usually affects over half of the choroid & enlarges very slowly. Presentation is usually in the 2nd decade despite the fact the tumour is present at birth. Complications include secondary retinal cystoid degen-

eration and exudative retinal detactoment. Neovascular glaucoma can result if exudative retinal detachment is not treated.

Ultrasonography (B-scan) shows diffuse choroidal thickening. The fundus has a diffuse deep-red "tomato ketchup" colour that is more marked at the posterior pole. Treatment of vision threatening cases involves low dose radiotherapy or Photo Dynamic Therapy.

Conclusion :

The main systemic features are angiomas which may involve face, meninges and brain. Nevus flammeus is a congenital facial angioma which roughly involves the area of distribution of the first and/or second division of the trigeminal nerve. The skin lesion may be associated with hypertrophy of the involved area of the face. An ipsilateral angioma of the meninges and brain is common and atrophy of the surrounding cerebral cortex may lead to variable degrees of mental handicap. The intracranial lesion frequently calcifies & it can be detected by plain radiographs (linear cortical calcifications which appear as double densities-train tract sign). Treatment should be multidisciplinary approach in collaboration with ophthalmologist, paediatric neurologist & dermatologist also.

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