Sturge Weber Syndrome-A 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, port wine stains, dilated ocular vessels and glaucoma[1].

Keywords- *angiomas, port wine stain, sturge Weber syndrome*

Introduction

Sturge-Weber syndrome belongs to a group of disorders known as the phakomatoses (mother-spot diseases)[1]. It consists of congenital hamartomatous malformation that may affect the eye, skin, and central nervous system at different times, characterised by the combination of venous angiomas of leptomeninges, face, jaws, oral soft tissue[3]. Sturge Weber syndrome was first described by Schirmer in 1860. More scientific description was given by Sturge in 1879.

Sturge Weber syndrome is believed to be caused by the persistence of vascular plexus around cephalic portion of the neural tube. This plexus develop during the sixth week of intrauterine development but normally undergoes regression during ninth week[3].

Angiomas of leptomeninges are usually unilateral, located in parietal and occipital region. The presence of angioma results in precipitation of calcium deposits in cerebral cortex underlying it[2]. Seizures, mental retardation, hemiplegia, or hemiparesis may be present, depending on the extent of lesion[4].

The cutaneous angiomas are called port wine stain which usually occur unilaterally along dermatomes supplied by the ophthalmic and maxillary division of trigeminal nerve. It may be bilateral or totally absent or may extend to neck, limbs, and other parts of the body. Involvement of the areas supplied by ophthalmic division is pathognomic[1]. Ocular involvement can result in glaucoma, choroidal hemangioma, buphthalmos .

Intraoral angiomatosis may involve lips , buckle mucosa, palate, gingival and floor of mouth. This syndrome is of rare occurrence and management becomes complicated due to risk of hemorrhage.

Case Report

A 5 months old female child brought to outpatient department of the department of pediatrics, with a chief complaint of convulsions of left half of body and weakness (decreased movements) of left side of body since 2 days.

She is first born of noncasanguinous marriage.

Birth history was normal.

Family history is noncontributory.

On Clinical examination, child had the port wine stain, a reddish colored macule present bilaterally over upper part of the face, involving ophthalmic and maxillary distribution of trigeminal nerve [Figure no.1].

A black colored nevus present on lower ,buttocks, back and lower limb(figure no.2).

Blue sclera present in both eyes [figure no.4].

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Neurological examination revealed reduced power with increased tone and brisk reflexes over left half of the body.

Child was further investigated in view of neurocutaneous marker and abnormal neurological examination.

1. CT Brain suggestive of intracranial calcification "rail road calcification" on right frontal lobe with atrophy of right frontal & partial lobe[figure no.3]

2.Ophthalmic evaluation of eyes revealed glaucoma in both eyes, retinal vessels are normal.[figure no.4]

Discussion

Sturge – Weber syndrome is sporadic vascular disorder consists of port wine stain (facial capillary malformation) leptomeningeal angioma (abnormal blood vessels of brain) and abnormal blood vessels of eye leading to glaucoma[2].

An estimated incidence of sturge- Weber syndrome is approximately 1 per 50,000 live births.

My patient 5 months old female child presented with "Port wine stain" on face, History of convulsion, hemiparesis involving left half of the body, glaucomatous changes present in both eyes, CT brain is suggestive of "rail road calcification" on right side.

Etiopathology

- Sturge - Weber syndrome is supposed to be due to somatic mutations but still has not been demonstrated[1].
- CNS changes appears to result due to chronic hypoxic state leading to cortical atrophy and calcifications[4].
- Facial Port-Wine stain is thought to be a result of anomalous development of embryonic vasculature in early stages of development[3].

Clinical Manifestation

The facial port wine stain is present at birth and tends to be unilateral ,sometimes may be bilateral and mostly

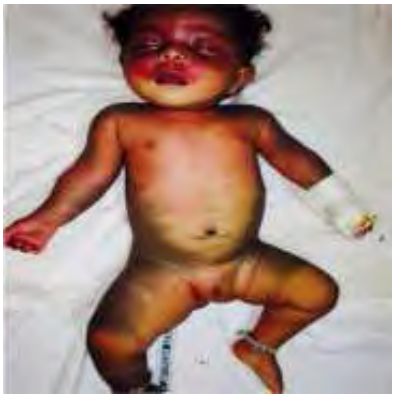

Figure no. 1 : clinical picture of the child showing port wine stain and Mongolian spot over lower back , buttocks and thighs

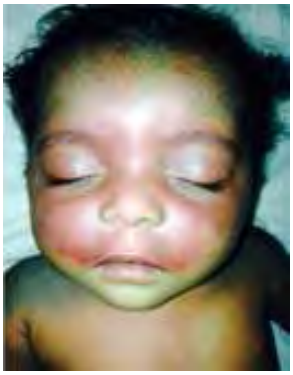

Figure no.2 : clinical picture of child showing bilateral facial port wine stain involving ophthalmic and maxillary branches of trigeminal nerve

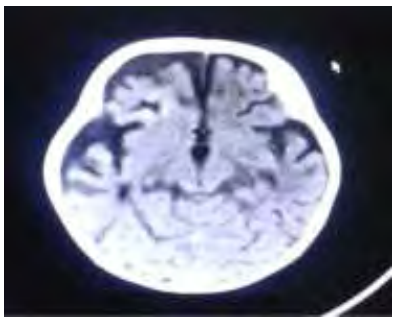

Figure no.3 : CT brain showing "rail road calcification" involving right frontal lobe with atrophy of right frontal and parital lobe

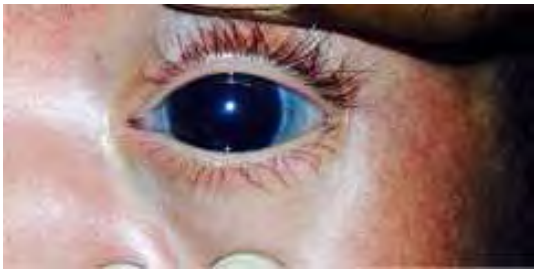

Figure no.4 : glaucomatous changes present , large cornea and blue sclera is visible.

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b) Books and Monograph :

i) Anemia, In: Cotran RS, Kumar V, Collins T. Robbins Pathologic Basis of Disease. 6th ed. Singapore. WB Saunders Company, 1999: 1300-1321.

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