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Sturge - Weber Syndrome - A Rare Case Report

Amala B

Department of Dermatology, Venerology & Leprosy,Madras Medical College and Government General Hospital

Abstract

SturgeWeber angiomatosis is a rare, nonhereditary developmental condition characterized by a hamartomatous vascular proliferation involving the tissues of brain and face. Here we report a case with port wine stain and central nervous system involvement.

Keywords : Angiomas, port wine stain, SturgeWeber syndrome

Introduction

Sturge-Weber syndrome or encephalotrigeminal angiomatosis is a rare congenital, non- familial, phakomatosis of unknown etiology, which occurs with a frequency of 1:50,000 live births.[1] It is characterized by capillary malformation located on the branches of the trigeminal nerve, homolateral leptomeningeal capillary- venous malformation and ocular abnormalities (glaucoma and choroidal hemangioma) . This malformation can cause epilepsy, cerebral atrophy and mental retardation.

Case report

A 2 year old female child born of third degree consanguineous marriage presented with complaints of red coloured skin lesion over right side of face and multiple dark coloured skin lesions over right upper limb, right side of chest, abdomen and both thighs since birth, which were progressively increasing in size. There was history of multiple episodes of seizures since 45th day of life. Seizure involved the left side of body and the duration of each episode was 3- 5mins. Child was on anti epileptic drugs since 2 months of age. Parents gave history of delayed developmental milestones.

There was no significant antenatal and family history.

On examination, child was active, port wine stain was seen over right side of face from forehead to cheek along the distribution of trigeminal nerve, right upperlimb, right side of chest, abdomen and both thighs. Ophthalmic examination and oral mucosa were normal. Diagnosis of Sturge- Weber syndrome was made.

Fig.1 : Port Wine Stain on right side of face

Fig. 2 : Port Wine Stain on right upper limb, chest and abdomen

MRI showed right cerebral atrophy with early gyral calcification and abnormal venous channels in the right frontoparietal region subarachinoid space with deformed superficial veins.

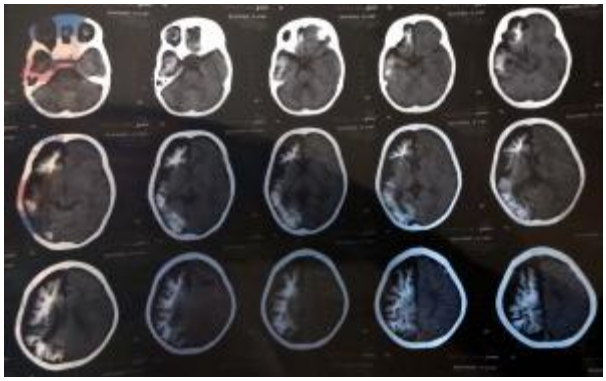

Fig.3 : Brain Radiography showing right cerebral atrophy and calcification

Blood investigations were normal. Based on history, clinical and radiological findings, diagnosis of Sturge-Weber Syndrome was confirmed. Child was advised to continue anti epileptic drugs with regular follow- up.

Discussion

Sturge-Weber syndrome (SWS) is a congenital but not inherited neurocutaneous disorder classically characterized by port-wine stain over the face, vascular eye abnormalities and ipsilateral leptomeningeal angioma. The leptomeningeal malformations lead to venous hypertension and subsequent hypoperfusion of the underlying cortex. Children with SWS often develop progressive problems like glaucoma, seizures, stroke, and intellectual disability. This syndrome occurs with equal frequency in both sexes [1, 2]

SWS is referred to as complete when both CNS and facial angiomas are present and incomplete when only one area is affected without the other. The Roach Scale used for classification, as follows: 1

Type I - Both facial and leptomeningeal angiomas; may have glaucoma
Type II - Facial angioma alone (no CNS involvement); may have glaucoma
Type III - Isolated leptomeningeal angioma; usually no glaucoma.

According to the above criteria, our case belongs to complete Type I SWS. The port wine stain is a congenital malformation of the dermis that involves venules, capillaries, and possibly perivenular nerves. It occurs in an estimated 3/1000 births.[1] It presents at birth and typically involving the distribution of Trigeminal nerve especially the ophthalmic division . It is usually unilateral but may be bilateral or totally absent or may extend to neck, limbs and other parts of the body.[1] This may also be associated with unilateral limb hypertrophy.[3]

Neurological deficit is caused by the intracranial angiomas. These are located typically on the occipital region of the brain on the same side as the port wine birthmark.

These angiomas create abnormal conditions for brain function in the region. Seizure activity is the most common early problem, often starting by one year of age, present in 55- 90% of cases.[1,2,6] Our patient developed seizures at the age of 2 months. The convulsions usually appear on the opposite side of the body and vary in severity. Developmental delay is not universal and is present in 50% of cases.[3,6] Imaging findings consist of cortical calcifications - tram line calcifications (30%), cortical atrophy, enlarged ipsilateral choroid plexus and pial angiomatosis.[1]

Eye manifestations include glaucoma (30-70%) and buphthalmos (up to 50% of newborns with SWS). [1,3] Glaucoma can be present at birth or develop later. The risk of glaucoma is highest in the first decade .The glaucoma is usually restricted to the eye involved. When it presents at birth, the eye is enlarged (Buphthalmos) and the cornea appears cloudy.3 In this present specific case, the child had normal ophthalmological examination.

Oral changes occur in 40% cases of this syndrome and may include massive growth of the gingival and asymmetric jaw growth. Infants affected with Sturge-Weber Syndrome need to be monitored by a pediatrician, neurologist, ophthalmologist and dermatologist.[4,5]

Laser treatment is available for port wine stain in children as young as one month of age. Anticonvulsants are used to control the seizures. Brain surgery may also find a role in seizure management. Eye drops and or oral medications are used to control the glaucoma.

Conclusion

Children with Sturge- Weber syndrome should be managed by a team of pediatrician, neurologist, ophthalmologist and dermatologist. Early diagnosis and appropriate treatment will help to minimize the morbidity**.**

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