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Original Research Paper

General Medicine

A RARE NON-HEREDITARY DEVELOPMENTAL DISORDER OF PHACOMATOSES

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ABSTRACT Sturge weber syndrome is a rare non-hereditary developmental condition, also called enchaphalotrigeminal angiomatosis characterized by a hamartomatous vascular proliferation involving the tissues of brain and face. We present here a case of an young boy presenting with recurrent tonic clonic seizures from 6 years associated with a port wine stain on left upper half of his face. His brain imaging findings are characteristic of Sturge Weber syndrome.

KEYWORDS :

INTRODUCTION

Sturge–Weber syndrome (SWS) belongs to a group of disorders collectively known as the phacomatoses ("mother-spot" diseases).It consists of congenital hamartomatous malformations that may affect the eye, skin, and central nervous system (CNS) at different times, characterized by the combination of venous angioma of leptomeninges, face, jaws and oral soft tissues. SWS was first described by Schirmer in1860. More specific description was given by Sturge in 1879. SWS is believed to be caused by the persistence of vascular plexus around the cephalic portion of the neural tube. This plexus develops during the sixth week of intra uterine development but normally undergoes regression during ninth week.

Case Report -

A 18 year old boy mechanic by occupation presented to casualty department with Status Epilepticus. He had history of recurrent generalized tonic clonic seizures and right sided weakness of upper and lower limbs since around 6 years. He has reddish discoloration of left half of forehead (port wine stain) since birth which gradually darkened with age. He had gum hypertrophy and history of past gingival bleeding. There is no history of fever, headache, vomiting, head trauma, dizziness or diminished vision. He was taking phenytoin therapy which he self-stopped recently. There was no significant family or personal history. CNS examination revealed slight diminished power in right upper and lower limbs-4/5.

Routine blood investigations were within normal limits. Brain imaging -MRI was done which showed features suggestive of cerebral hemiatrophy with gyriform calcification which is characteristic of Sturge Weber syndrome.

Treatment and prognosis depends upon the nature and severity of clinical features. Presence of port wine stain can cause deep psychological trauma to patient and development of personality is affected in almost all patients. Port wine stains can be improved by dermabrasion, tattooing, and flash lamp pulsed dye lasers. He was on phenytoin from outside, which he stopped since last 2 months. At admission he was given loading dose of Levetiracetam and continued on maintenance dose. Physical therapy advised for paralysis or weakness and Psychological counseling of parents were done.

DISCUSSION

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 is used for classification, as follows

Type I - Both facial and leptomeningeal angioma; may have glaucoma

Type II - Facial angioma alone (no CNS involvement); may have glaucoma

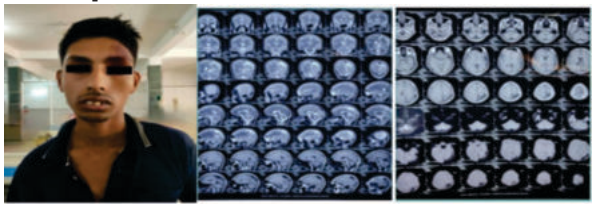
Type III - Isolated leptomeningeal angioma; usually no glaucoma.

According to the above criteria, our case is complete Type I SWS case.

The differential diagnosis included Rendu-

Osler-Weber syndrome, Angio-osteodystrophy syndrome, Maffucci syndrome and Klippel-Trenaunay-Weber syndrome. This is a case of recurrent seizure with unilateral weakness of limbs with gingival hypertrophy with port wine stain thus diagnosed as Sturge Weber syndrome(a rare syndrome) based on clinical and radiological grounds.

Sturge-Weber syndrome presents with a large number of clinical manifestations and early diagnosis is difficult. Early diagnosis is necessary to avoid future complications. Expert psychological counselling of patients and their parents is necessary.



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