

Sturge-Weber Syndrome: Rare Neurocutaneous Disorder

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Sturge-Weber syndrome (SWS) is a rare neurocutaneous disorder which occurs sporadically. It belongs to a group of disorders collectively known as phakomatoses and consists of congenital hamartomatous malformations [1]. It is a congenital but non-hereditary disease. SWS presents with hamartomatous malformations, which results due to the failure of fetal veins to develop normally in the brain, skin and eye. This leads to venous hypertension and subsequent hypoperfusion of the underlying cortex resulting in chronic cerebral ischemia, atrophy and neurological deterioration.

It is characterized by a birth mark called port wine nevi, associated with abnormalities of the brain caused by abnormal blood vessels (angiomas) that occur on the cerebral cortex [2, 3]. These changes are usually unilateral. Bilateral involvement is seen in only 15% cases [4]. Port wine nevi are congenital malformations in the dermis of the skin involving venules, capillaries and possibly perivenular nerves [5]. It is also associated with choroidal vascular lesions, glaucoma, seizures, neurologic deterioration and eventual neurodevelopmental delay.

SWS occur with equal frequency in both sexes, with seizures typically developing in the first year of life [6] with a frequency of 1:50,000 [13]. Neurological deficit is caused by the vascular malformation within the intracranial vessels [7]. Glaucoma and vascular malformations involving conjunctiva, episclera, choroid and retina are the ocular manifestations [8].

Plain radiographs of the skull show confluent "tram-track" calcifications. Computed tomography (CT) scan shows the gyriform calcifications with cortical atrophy, enlarged and enhancing ipsilateral choroid plexus and angiomas [9]. Cortical calcifications present at birth are reported in 30% of the cases [10]. Magnetic resonance imaging (MRI) is the best imaging modality for diagnosing SWS while calcifications can be assessed in detail on CT images.

If the onset of seizure is before the age of 2 years, it has poor prognosis with mental retardation and refractory epilepsy [9]. SWS has been reported in neonates with seizures in about 75% to 90% of

patients [11]; even a case of 2 days old baby with SWS and seizure has been reported in literature [10]. Portwine stain is common in children. According to Enjolras et al, patients with lesions located in the ophthalmic and trigeminal distribution areas are at risk for associated neuro-ocular symptoms [11]. Most cases with SWS are not life threatening; however, it is a progressive disease associated with continuous neurological deterioration [9].

SWS can be classified into three types; type I - both facial and leptomeningeal angiomas (may have glaucoma), type II - facial angiomas alone (may have glaucoma) and type III - isolated leptomeningeal angiomas (usually no glaucoma) [12].

There is a vast list of the differential diagnosis of SWS which includes Klippel Trenaunay Weber syndrome, Rendu-Osler-Weber syndrome, Bannayan Riley Ruvalcaba syndrome, Divry Van Bogart syndrome and Cobb syndrome; however, the exact differentiation among these entities is not always possible due to overlapping features in many of these syndromes [10].

Management of this syndrome involves both medical and surgical approaches. Medical treatment includes anticonvulsant therapy with prophylactic low dose aspirin to prevent thrombus formation. Port wine stains on the face can be treated by cosmetic surgeons, laser therapy or dermabrasion [13]. SWS is diagnosed and treated on the basis of history, clinical examination and imaging modalities along with the co-ordination among different fields of medicine, surgery and dentistry. It is difficult to determine the exact etiology of SWS but further research in this field can help in understanding the disease and application of new treatment options.

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