

Sturge Weber Syndrome : A Review

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Abstract

Sturge Weber Syndrome also called as encephalotrigeminal angiomas is a sporadically occurring neurocutaneous syndrome caused by persistence of transitory primordial arteriovenous connection of the foetal intracranial vasculature. It is characterized by vascular malformation with capillary venous angiomas that involve face, choroid of eye and leptomeninges with resulting neurological and orbital manifestations. The diagnosis is usually considered in a child presenting with seizures and facial capillary malformation along the trigeminal nerve distribution. The characteristic imaging features on computed tomography and magnetic resonance imaging lead to the diagnosis. Treatment mainly consists of seizure control with anticonvulsants. Early and effective control of seizures markedly improves the prognosis. Surgery is reserved for refractory seizures, intractable glaucoma and laser therapy for facial naevus flammeus. However subnormal intelligence and neurological deficits are inevitable in most patients.

Introduction

Sturge Weber syndrome was first described by Sturge in 1879 in a six year old girl with facial naevus and Kalischer in 1897 demonstrated the cerebral involvement.¹ The diagnosis of Sturge Weber syndrome is considered in a child with facial naevus flammeus and seizure but in the absence of cutaneous manifestations the diagnosis remains elusive. Radiological investigations are most useful in such circumstances with computed tomography and magnetic resonance imaging playing a pivotal role in demonstrating the cerebral changes. Once the diagnosis is made early and effective seizure control is mandatory for an improved prognostic outcome.

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Pathophysiology

Angiomas of Sturge Weber syndrome result due to failure of regression of a vascular plexus around cephalic portion of neural tube which is destined to become facial skin. This vascular plexus normally forms at 6th week of intrauterine life and regresses by 9th week. Failure of its regression results in residual vascular tissue which forms angiomas of leptomeninges, face and ipsilateral eye.² These blood vessels show abnormal blood flow pattern as vasomotor phenomenon, venous occlusion, thrombosis and “vascular steal phenomenon” resulting in ischaemia, gliosis, atrophy and calcification of underlying cortical tissue.² Although the leptomeningeal angioma in Sturge Weber syndrome is typically a static lesion, it has been demonstrated by some to be of progressive nature.³

The main ocular manifestations — buphthalmos and glaucoma occur due to secondary increase in intraocular tension due

to increased secretion of aqueous humor by choroidal haemangioma.

Clinical Manifestations

Sturge Weber syndrome is a neurocutaneous syndrome (phakomatosis) with a sporadic occurrence. Its exact incidence is not known.⁴ It has no racial or sex predilection. The typical patient presents at birth with facial angiomas, however the reverse is not always true. In the incomplete form of Sturge Weber syndrome, CNS angiomas occur without cutaneous manifestation thus no suspicion of the syndrome arises until the onset of seizures.

Clinical features

- a) Seizures: Epilepsy is a very common feature and often occurs during the first year of life.⁵ Seizures result from cortical irritability caused by angioma through the mechanism of hypoxia, ischaemia and gliosis. About 80% of affected persons have focal seizures involving the contralateral side of the portwine stain.⁶ The hypoperfusion of cortical tissue is further accelerated by seizures, thereby worsening the prognosis.⁷
- b) Developmental delay and mental retardation are almost always associated with seizures. Other neurological manifestations include headache or hemiparesis.
- c) Facial naevus flammeus (Portwine stain): Congenital macular lesions which may be initially light pink turning dark purple and thicker later on. Portwine stain involves the area of distribution of trigeminal nerve. Those with involvement of entire first division of trigeminal nerve have highest risk of associated Sturge Weber syndrome.⁸ It is to be emphasized that the syndrome may occur without facial naevus and not all

patients with port wine stain have Sturge Weber syndrome.

- d) Glaucoma and buphthalmos typically occur when portwine stain involves eyelids. It can develop at any age, is usually unilateral and ipsilateral to portwine stain. It occurs due to increased secretion of aqueous humor by choroidal haemangioma.
- e) Other features include visual loss, macrocephaly and hemiatrophy. Soft tissue hypertrophy occurs in patients who have combined features of Klippel Trenaunay Weber syndrome.⁹

Diagnosis

The diagnosis of Sturge Weber syndrome is based on imaging studies, although CSF analysis may reveal elevated protein due to micro haemorrhages. Skull films may reveal tram track calcification caused by calcification in apposing gyri, ipsilateral calvarial thickening and enlargement of the paranasal sinuses and mastoid. Cranial CT demonstrates abnormal contrast enhancement of angioma, enlarged choroid plexus ipsilateral to the angioma and abnormal draining medullary and subependymal veins (Fig. 1). Cortical atrophy underlying the angioma with gyriform "tram track" calcification is the characteristic imaging feature. Calcification however is unusual before 2 years of age and most commonly involves the parietal and occipital lobes (Fig. 2). MRI is the current "gold standard" for diagnosis of disease which is reliable even in very young infants.¹⁰ MRI demonstrates thickened cortex, decreased convolutions, abnormal white matter and gadolinium enhancement of leptomeningeal angioma. Sinovenous occlusion can be seen on MR Venography. Angiography reveals lack of superficial cortical veins and nonfilling of

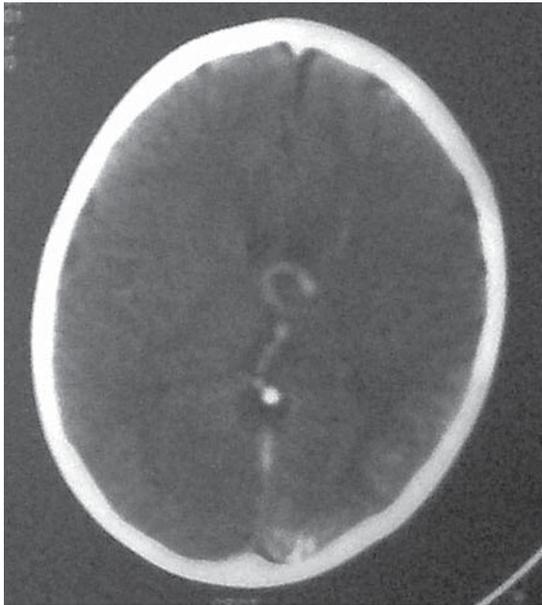


Fig. 1 : Postcontrast CT image in a patient with Sturge Weber syndrome showing abnormal enlarged subependymal vein.

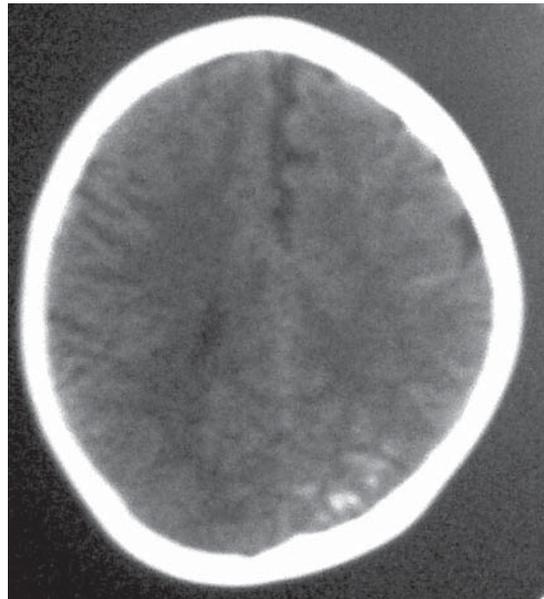


Fig. 2 : Precontrast CT scan showing gyral calcification in left parietooccipital cortex in the same patient.

dural venous sinuses with abnormal tortuous vessels that course towards the vein of Galen. Other vascular abnormalities demonstrated on angiography include thrombotic lesions and arteriovenous malformations.¹¹

Functional cerebral studies using PET and SPECT have shown abnormalities of metabolism and perfusion in Sturge Weber syndrome.¹² A study of cerebral blood flow using SPECT in very young infants with Sturge Weber syndrome by Pinton *et al*⁷ revealed a specific functional pattern before the onset of seizures. During the first months of life, the vascular malformation tends to be hyperperfused. The pattern switches to hypoperfusion at the end of the first year, accelerated by seizures and due to progressive ischaemia in the parenchyma underlying the vascular malformation. They suggested that seizures are responsible for decreased flow of blood to underlying cerebral cortex and thus epilepsy acts as an additional worsening

factor in Sturge Weber syndrome.

Differentiation of Sturge Weber syndrome from other varieties of arteriovenous malformation as Klippel Trenaunay Weber syndrome, Rendu-Osler Weber syndrome, Bannayan Riley Ruvalcaba syndrome, Divry Van Bogart syndrome and Cobb syndrome is important. However exact categorization of the lesions is not always possible due to overlapping features in many of these syndromes.

Treatment

Medical management of Sturge Weber Syndrome includes treatment for seizures. Seizures are typically focal thus an anticonvulsant with efficacy against focal seizures is preferable. The goal of treatment for glaucoma is to prevent optic nerve atrophy. Carbonic anhydrase inhibitors, β -antagonists are used to control intraocular pressure.

Aspirin has been used for headache and to prevent vascular disease. It has to be used with extreme caution in children due to concern about Reye syndrome. Thomas Sohl *et al*¹³ reported a decreased incidence of stroke like events in patients who received aspirin.

Portwine stain requires treatment with pulsed tunable dye laser which should be started as soon as possible. Patients with rapidly progressive convulsive disorders that are recalcitrant to treatment should be considered for surgical extirpation of the affected lobe or hemispherectomy. Other surgical options include corpus callosotomy and vagal nerve stimulation. Early surgery has been advocated for better seizure control, to improve outcome and prevent developmental delay.¹⁴

Prognosis

Seizures play a major role in failure of mental development and deterioration of mental function, hence effective seizure control is omnipotent.

Factors associated with poor prognosis are:

1. Early onset seizures.
2. Seizures refractory to medical treatment
3. Extensive leptomeningeal angioma
4. Extensive atrophy of underlying cerebral cortex
5. Development of hemiparesis and
6. Deterioration in cognitive functioning

To conclude, the diagnosis and management of patients with Sturge Weber syndrome requires the combined skills of a physician, radiologist and a psychologist.

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