

# Unusual Presentation of Sturge Weber's Syndrome with Cataract : A Case Report

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## Abstract

A 6 yr old girl came to our OPD with a decreased vision in the right eye since 2 yrs. She had a portwine stain over her right forehead since birth not decreasing in size or intensity. Her vision in the right eye was HM and that in the left eye was 6/6. On examination of the anterior segment, she had a mature cataract in her right eye. Her left eye was normal. Gonioscopic and indirect ophthalmoscopic examination detected no abnormality. Her CT scan brain was also normal. Her paediatric opinion was taken which confirmed the diagnosis of Sturge-Weber syndrome.

## Introduction

The Sturge-Weber syndrome (SWS), also called encephalotrigeminal angiomatosis, is a neurocutaneous disorder with angiomas involving the leptomeninges (leptomeningeal angiomas) and skin of the face, typically in the ophthalmic (V1) and maxillary (V2) distributions of the trigeminal nerve. The cutaneous angioma is called a port-wine stain (PWS).

In the brain, LAs demonstrated by structural neuroimaging may be unilateral or bilateral; unilateral angiomas are more common.

The neurologic manifestations vary, depending on the location of the LAs, which most commonly are located in the parietal and occipital regions, and the secondary effects of the angioma. These include seizures, which may be intractable; focal deficits, such as hemiparesis and hemianopsia, both of which may be transient, called "strokelike episodes"; headaches; and developmental disorders, including developmental delay, learning disorders, and

mental retardation. Developmental disorders are more common when angiomas are bilateral.

The primary complications involving the ipsilateral eye are buphthalmos and glaucoma, with treatment aimed at controlling the intraocular pressure (IOP) and preventing progressive visual loss and blindness. Cosmetic concerns are also important, and laser therapy is available for the PWS. Extracranial angiomas and soft-tissue overgrowth also may occur. Certain CNS malformations have been associated with the syndrome; other neurocutaneous disorders are included in the differential diagnosis.

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

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

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### Case Report

A 6 yr old girl came to our OPD with a decreased vision in the right eye since 2 yrs. She had a portwine stain over her right forehead since birth not decreasing in size or intensity. Her vision in the right eye was HM and that in the left eye was 6/6. on examination of the anterior segment she had a mature cataract in her right eye. Her left eye was normal. Gonioscopic and indirect ophthalmoscopic examination detected no abnormality. Her CT scan brain was also normal. Her paediatric opinion was taken which confirmed the diagnosis of Sturge-Weber Syndrome.

She was operated for right eye cataract surgery under general anaesthesia with intraocular lens implantation (Figs. 1 and 2).

### Pathophysiology

SWS is caused by residual embryonal blood vessels and their secondary effects on surrounding brain



Fig. 1



Fig. 2

tissue. A vascular plexus develops around the cephalic portion of the neural tube, under ectoderm destined to become facial skin. Normally, this vascular plexus forms in the sixth week and regresses around the ninth week of gestation. Failure of this normal regression results in residual vascular tissue, which forms the angiomas of the leptomeninges, face, and ipsilateral eye.

The main ocular manifestations (i.e., buphthalmos, glaucoma) occur secondary to increased IOP with mechanical obstruction of the angle of the eye, elevated episcleral venous pressure, or increased secretion of aqueous fluid.

### Frequency

**In the US :** The incidence of SWS is estimated at 1 per 50,000. No regional differences have been identified. The inheritance is sporadic. The incidences of the major clinical manifestations of SWS are listed in Table 1.

### Mortality/Morbidity

Neurologic and developmental morbidity includes seizures, weakness, strokes, headaches, hemianopsia, mental retardation, and developmental abnormalities. The development of seizures and the age of onset may correlate with the degree of neurologic involvement. Neurologic dysfunction increases with bilateral PWS. Patients may experience complications related to refractory seizures and anticonvulsants, visual loss and blindness from glaucoma, cosmetic deformities, and other manifestations of soft-tissue involvement.

### Race

No racial differences have been reported.

**Table 1 : Clinical Manifestations of Sturge-Weber syndrome**

Risk of SWS with facial PWS	8%
SWS without facial nevus	13%
Bilateral cerebral involvement	15%
Seizures	72-93%
Hemiparesis	25-56%
Hemianopia	44%
Headaches	44-62%
Developmental delay and mental retardation	50-75%
Glaucoma	30-71%
Choroidal haemangioma	40%

## Sex

Both sexes are affected equally.

## Age

The typical patient presents at birth with facial angiomas; however, not all children with facial angiomas and PWS have SWS, which raises certain diagnostic and prognostic concerns.

In the "incomplete" forms of SWS, CNS angiomas occur without cutaneous features (Type III, Roach Scale), and therefore, no suspicion of SWS arises until a seizure or other neurologic problem develops. Thus, the diagnosis of SWS is not always straightforward.

## History

The Sturge-Weber Foundation maintains a list of patients with SWS; its efforts have promoted clinical and scientific research, which have led to improvement in the treatment of SWS. This includes studies on the natural history of the disorder.

- Facial nevus, PWS
  - These are congenital macular lesions that can be progressive; they may be a light pink colour initially and then progress to a dark red or purple nodular lesion. These may be isolated to the skin, associated with lesions in the choroidal vessels of the eye or the leptomeningeal vessels of the brain, or even located on other body areas. A PWS may be difficult to visualize in a patient with dark skin pigmentation.
  - Not all people with a PWS have SWS; the overall incidence of SWS has been reported to be 8-33% in those with a PWS.
- Seizures, refractory seizures
  - The incidence of epilepsy in patients with SWS is 75-90%; seizures may be intractable. Seizures result from cortical irritability caused by cerebral angioma, through mechanisms of hypoxia, ischaemia, and gliosis.
  - Dual pathology, such as microgyria, also may be present, which also contribute to epileptogenesis.
- Focal versus generalized seizures
  - Since the lesion responsible for the epilepsy in SWS is focal, the majority of seizures are focal seizures.
- Status epilepticus
  - Prolonged seizures cause neurologic injury secondary to metabolic disturbances such as

hypoxaemia, hypoglycaemia, hypotension, ischaemia, and hyperthermia.

- In an already compromised vascular system, such as a vascular steal from the angioma, seizures are more likely to cause injury, even when short. Episodes of status epilepticus are, therefore, especially dangerous in SWS.

- Hemiparesis: The incidence is approximately 33%, varying from 25-56%; it occurs secondary to ischaemia with venous occlusion and thrombosis. Commonly, transient weakness may occur with seizures and may increase with recurrent seizures. Transient hemiplegia may be accompanied by migraine headache, suggesting a vascular mechanism.
- Strokelike episodes: Transient episodes are referred to as strokelike episodes.
- Hemianopsia: The mechanism is similar to that of hemiparesis and is dependent on the location of the lesions.
- Developmental delay and mental retardation
  - These are related to the degree of neurologic involvement, occurring in 50-60% of patients; they are more likely in patients with bilateral involvement.
  - Seizures are associated with a higher incidence of mental retardation, and regression also may be related to the frequency and severity of seizures.
- Headaches
  - These occur secondary to vascular disease, giving symptoms of a migraine headache, considered "symptomatic migraine."
- Ocular manifestations, glaucoma and blindness
  - Glaucoma typically occurs in SWS only when the PWS involves the eyelids. The incidence ranges from 30-71%. Glaucoma may be present at birth but can develop at any age, even in adults.
  - Treatment includes yearly examinations, looking for optic nerve damage (with measurement of IOP and visual fields) and corneal diameter and refractive changes in children.
  - Glaucoma usually occurs only with an ipsilateral facial PWS, although it may be bilateral when facial involvement is bilateral. Contralateral glaucoma may develop, although rarely. Glaucoma also may occur without

neurologic involvement (Type II, Roach Scale).  
 - Glaucoma in SWS is produced by mechanical obstruction of the angle of the eye, elevated episcleral venous pressure, or hypersecretion of fluid by either the choroidal haemangioma or ciliary body. The anterior chamber angle abnormality is consistently seen in the infantile glaucoma cases in SWS, while increased episcleral venous pressure may have a key role in late-onset glaucoma cases in SWS. Decreased vision and blindness result from untreated glaucoma, with increased IOP leading to optic nerve damage. An acceptable range of IOP is 10-22 mm Hg.

- Buphthalmos (hydrophthalmia): Enlargement of the eye occurs from the same mechanisms as glaucoma.
- PWS, cosmetic problems, soft-tissue hypertrophy

#### Physical

- PWS
- Macrocephaly
- Eye - Buphthalmos, heterochromia of iris, tomato-catsup colour of the fundus (ipsilateral to the nevus flammeus) with glaucoma, possibility of choroidal angioma visible with an ophthalmoscope
- Soft-tissue hypertrophy
- Neurologic signs
  - Developmental delay/mental retardation
  - Learning problems
  - Attention deficit hyperactivity disorder
  - Hemiparesis
  - Visual loss
  - Hemianopsia

#### Lab Studies

Cerebrospinal fluid (CSF) protein may be elevated, presumably secondary to microhaemorrhage. Note that a major intracranial haemorrhage itself is rare in SWS, although microhaemorrhage may be common.

#### Imaging Studies (Table 2)

Endocrinologic tests : The Sturge-Weber Foundation notes increasing use of growth hormone in its members. Some have developed a body habitus similar to that in Cushing syndrome. This has occurred around the time of puberty.

**Table 2 : Summary of Work-up Findings in Sturge-Weber Syndrome**

CSF analysis	Elevated protein
Skull X-ray	Tram-track calcifications
Angiography	Lack of superficial cortical veins Nonfilling dural sinuses Abnormal, tortuous vessels
CT scan	Calcifications, tram-track calcifications Cortical atrophy Abnormal draining veins Enlarged choroid plexus Blood-brain barrier breakdown (during seizures) Contrast enhancement
MRI	Gadolinium enhancement of LA Enlarged choroid plexus Sinovenous occlusion Cortical atrophy Accelerated myelination
SPECT	Hyperperfusion, early Hypoperfusion, late
PET	Hypometabolism
EEG	Reduced background activity Polymorphic delta activity Epileptiform features

#### Histologic Findings

The leptomeninges appear thickened and discoloured by the LA, which fills the subarachnoid space, and abnormal venous structures are seen. Biopsies typically are not performed in SWS. However, pathologic specimens, such as those examined by Norman and Schoene, show calcium deposits in the cerebral vessel walls, in perivascular tissue and, rarely, within neurons, and neuronal loss and gliosis occur. These pathologic abnormalities may occur at a distance from the actual vascular lesion.

In skin biopsies of the port-wine stain in SWS, dilated ecstatic thin-walled vessels are seen in the superficial vascular plexus, but with no increase in the number of blood vessels.

In trabeculectomy specimens in patients with SWS, abnormal collagen depositions and abundant vessels in the intra-trabecular spaces have been seen

morphological abnormalities in the Schlemm canal. Haemangiomas in the trabecular meshwork are characteristic of SWS.

#### Medical Care

This includes anticonvulsants for seizure control, symptomatic and prophylactic therapy for headache, glaucoma treatment to reduce the IOP, and laser therapy for PWS.

- Seizures : Since the seizures are typically focal, an anticonvulsant with efficacy in focal seizures is preferable (see anticonvulsant medications).
- Glaucoma : The goal of treatment is control of IOP to prevent optic nerve injury (please see the articles on glaucoma in eMedicine Ophthalmology journal). Medications either decrease the production of aqueous fluid or promote the outflow of aqueous fluid. Beta-antagonist eye drops reduce the production of aqueous fluid, adrenergic eye drops and miotic eye drops reduce IOP by promoting drainage, and carbonic anhydrase inhibitors decrease IOP by decreasing production of aqueous fluid.
- Headaches : Recurrent headaches can be treated with symptomatic and prophylactic medications.
- Stroke-like events: Aspirin 3-5 mg/kg/day has been used for headaches and to prevent vascular disease, although it typically is used in patients who have had neurological progression or recurrent vascular events. Aspirin use needs to be with extreme caution in children, because of the concern about Reye syndrome, and the risks and benefits need to be carefully weighed.
- PWS : These need to be evaluated within the first week of life and differentiated from haemangioma.
  - PWS are treated with laser therapy, which should start as soon as possible, since multiple treatments are needed and earlier treatment may reduce the number needed. Also, the smaller the lesion initially, the fewer the laser flashes needed to remove the lesion.

#### Surgical Care

Surgery is desirable for refractory seizures, glaucoma, and specific problems related to various associated disorders, such as scoliosis.

- Seizures, refractory seizures
  - Surgical options are available for seizures refractory to medical treatment, especially for

focal seizures. Surgical procedures include focal cortical resection, hemispherectomy, corpus callosotomy, and recently, vagal nerve stimulation (VNS).

- Glaucoma surgery : If medications are unable to lower IOP, surgery may be beneficial. Trabeculectomy increases the release of aqueous fluid from the anterior chamber and opens the outflow pathway. Goniotomy is similar but is done through the eye. A Molteno valve can be placed (similar to a shunt), and cyclodestructive procedures with either freezing or laser decrease the production of aqueous fluid.

#### Consultations

Primary-care providers should be educated about SWS. Consultations are needed from a neurologist, an epileptologist (especially if seizures are intractable), a dermatologist, a plastic surgeon, a psychologist, a psychiatrist, a neuropsychologist, and a neuroendocrinologist.

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Another limitation is that internet use is a sedentary behaviour. Could time on the internet be better spent exercising or consuming vegetables? Perhaps limiting time on the internet to allow for exercise and healthy eating is an appropriate compromise.

**Beth A Lewis, The Lancet, 2007; 370 : 1891-92.**