# Sturge-Weber Syndrome

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Sturge-Weber syndrome (SWS) is one of the most common neurocutaneous syndromes or congenital phakomatoses. Encephalotrigeminal angiomatosis also refers to SWS since the hallmark vascular lesions involve the areas innervated by the trigeminal nerve and ipsilateral brain and meninges.[1] Therefore, it is a disorder of the vasculature of the face, meninges, brain, and eye. Unlike most other phakomatoses, SWS does not have a familial, racial, or gender predilection.[2] In 1879, Sturge[3] suggested a correlation between the cutaneous port-wine stain and contralateral neurological deficits in describing a child with both of these findings. Frederick Parkes Weber,[4] a British physician after whom many diseases are named, was the first to radiographically demonstrate cortical calcifications in SWS patients.

## Definition

Sturge-Weber syndrome is defined by a facial port-wine nevus and ipsilateral leptomeningeal angiomatosis.[2] Patients with SWS typically present with a combination of cutaneous angiomatosis, epileptic seizures, hemiplegia, visual field defects, and glaucoma, with the facial nevus flammeus often being the first apparent feature of the syndome.[5] The full syndrome implies cutaneous and cerebral findings, however, some patients lack one of these two.

Nevus flammeus presents most often as a transient salmon patch and less frequently as a persistent port-wine stain.[6] These two types differ in anatomical location and clinical course; the salmon patch is the midline nevus flammeus and the port-wine stain is the lateral nevus flammeus. The nevus flammeus is a vascular ectasia, not a hemangioma because it has no endothelial proliferation.[6]

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Figure not available online

**FIGURE 1.** A child with Sturge-Weber syndrome with a port-wine stain on the right side of his face (courtesy of the Sturge-Weber Foundation).

## Clinical Characteristics

In a study of 30 children with SWS, the natural history of the clinical features and various types of SWS are well summarized.[7] Twenty-eight had typical facial port-wine cutaneous angiomas, of which 19 had ipsilateral leptomeningeal angiomatosis (SWS type I). Nine had no intracranial lesions on computerized tomography (CT) (SWS type II). Nine presented with glaucoma at birth, while 15 developed glaucoma later. Fourteen had normal IQs, 14 had mild intellectual impairment, and only 2 had moderate intellectual impairment. Those with extensive brain involvement had refractory epilepsy and mental retardation. Seizures were noted in 22 patients, 18 of whom experienced their first epileptic seizure before 8 months of age. Twenty children had typical leptomeningeal

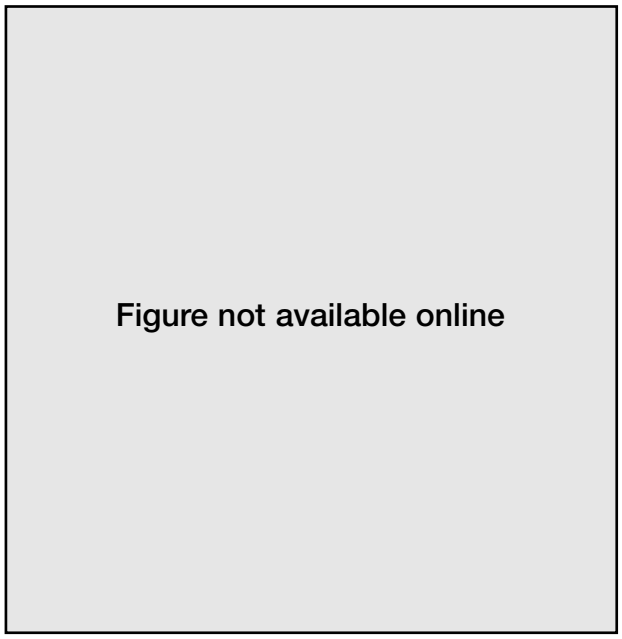


**FIGURE 2.** A 2 year old child with a port-wine stain on the left side of her face (courtesy of the Sturge-Weber Foundation).

angiomas, of which 18 had gyriform calcifications. Electroencephalogram changes of decreased voltage were seen in 26 of the patients.

Port-wine stains occur in about 0.3% of live births.[5,8] Facial port-wine stains tend to be distributed unilaterally and away from the midline of the face, although bilateral distribution and extension to the trunk, neck, lips, and oral mucous membranes can occur (Figures 1 and 2). The port-wine stains of SWS most often involve the forehead and upper eyelid, which are innervated by the first and second branches of the trigeminal nerve.[9] They vary in size from a few millimeters to several centimeters in diameter, are irregularly shaped, and grow commensurate with the child by thickening and changing color from pink to red to a deep purple.[10] The nevus flammeus may be adjacent to a nevus anemicus.[11]

The neurological findings of SWS are usually first evident within 2 years of birth. Seizures, which are often associated with a febrile illness, are the most common neurological manifestations in infants with SWS.[9] Initially, focal motor or generalized tonic-clonic seizures predominate. As the disease progresses, the patients may also have complex-partial seizures.[2] In addition, some patients have focal neurological defects, including unilateral hemiparesis or hemiplegia, a visual field disturbance, or intellectual impairment due to a contralateral angioma of the pia mater overlying a particular cerebral lobe.

Ocular defects of SWS include glaucoma, hemangiomas of the choroid, conjunctiva, and episclera, retinal detachment, vascular tortuosity, and dilatation and strabismus. Glaucoma, found in 30 to 50% of SWS, is almost always ipsilateral with a port-wine stain, which usually involves the upper eyelid, although involvement of both eyelids is common.[12,13] Choroidal hemangioma is another source of morbidity in SWS. Retinal degeneration overlying the choroidal tumor and continuous exudation of the hemangioma can result in visual field loss or retinal and choroidal detachment and require surgical intervention.[14] Fundoscopy may reveal a bright red "tomato catsup" fundus as a result of choroidal hemangiomas, which can be unilateral or bilateral.[15] Both enhanced CT and magnetic resonance imaging (MRI) can show diffuse choroidal hemangiomas in patients with SWS, but MRI is more sensitive and the preferred method for choroidal hemangioma detection.[14]

## Pathophysiology

The port-wine stain is a benign, congenital lesion that results from ectasia of cutaneous venules. During infancy, the superficial vascular plexus progressively dilates, without any of the endothelial proliferation that occurs in true hemangiomas.[10] Antibodies to S-100 protein demonstrate a decrease in the number of neurons surrounding the superficial venules, possibly resulting in decreased vascular tone and subsequent progressive dilation of the cutaneous superficial vascular plexi.[16] This nerve loss, which is probably of sympathetic autonomic origin, may be the pathological basis of the progressive vascular ectasias of SWS.[17]

The neuropathology of SWS is characterized by leptomeningeal and choroid plexus angiomatosis, and venous engorgement and tortuosity. The leptomeningeal angioma is believed to be the primary neuropathology of SWS. These angiomas consist of thin-walled veins within the pia mater of the posterior parietal, temporal, and anterior occipital lobes, and cause the meninges to become thicker and darker due to increased vascularity.[18] Using positron emission tomography and single photon emission computed tomography, studies of infants with SWS have shown hyperfusion of the region of the cortex involved in the vascular malformations during the first year of life, before the onset of seizures. Classic hypoperfusion and hypometabolism appear after 1 year of age (Figure 3).[19] These abnormalities of cerebral perfusion and glucose metabolism may be associated with neurologic deterioration and explain the increased prevalence of developmental delays in patients with SWS.[20,21] Furthermore, the progressive "railroad track" pattern calcification of the underlying cerebral cortex, which occasionally extends into the white matter, may also be evident.[18]

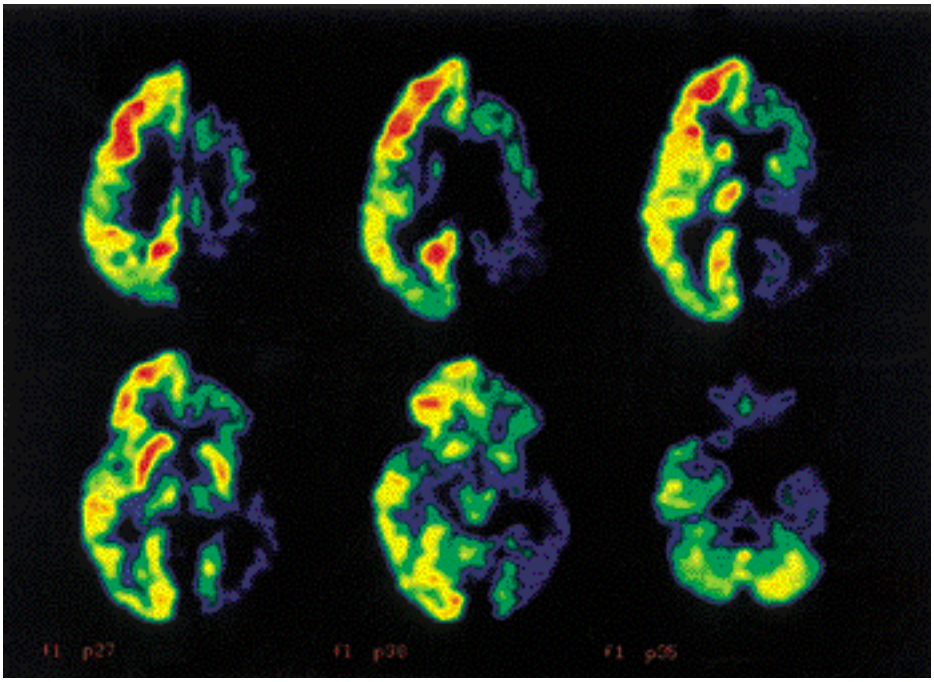

**FIGURE 3**. Positron Emission Tomography (PET) Scan from a patient with Sturge-Weber syndrome. Note the left side of the brain shows minimal glucose metabolism as compared with the right (courtesy of Harry Chugani MD, Professor of Pediatrics, Neurology and Radiology, Wayne State University School of Medicine and Director of the PET Center, Children's Hospital of Michigan).

## Differential Diagnosis

The nevus flammeus should be distinguished from other cutaneous vascular conditions such as the capillary hemangioma. Capillary hemangiomas are typically first evident shortly after birth or during infancy on the head or neck and tend to resolve by 12 years of age.[6] Furthermore, the port-wine stain (lateral nevus flammeus) is differentiated from the salmon patch (midline nevus flammeus) by location. Multiple vascular anomalies, sometimes with meningeal involvement may resemble SWS.[22] The correct diagnosis is important to identifying other clinical manifestations and treatment strategies.

## Treatment

Port-wine stains do not fade spontaneously. Treatments including cosmetic tattooing, surgery, grafting, cryotherapy, and radiation have been largely superseded by laser surgery.[23,24] This most recent treatment modality includes the flashlamp-pumped pulsed dye laser, copper vapor laser, argon ion-pumped dye laser, and neodymium yttrium aluminum garnet laser.[25] These lasers follow the theory of selective photothermolysis, in which the yellow laser light is preferentially absorbed by red blood cell oxyhemoglobin, which rapidly heats the red cells and causes coagulation and subsequent destruction of the superficial vessels, while sparing the surrounding tissue of the dermis and overlying epidermis.[26] The end result is destruction of the port-wine stain with minimal scarring.

Treatment of neurological dysfunction in SWS is focused on controlling seizures, because the majority of SWS morbidity results from complications of epilepsy.[27] Surgery can be used when seizures are unresponsive to medical treatment and, therefore, possibly related to arteriovenous malformations in the brain.[28] Surgical options include hemispherectomy, limited lobectomy, and corpus callostomy.

Ocular manifestations of SWS are also managed both medically and surgically. External beam irradiation and scattered photocoagulation have both been used to treat choroidal hemangiomas. Also, recent studies have demonstrated that glaucomatous complications are decreased when trabeculotomy is used in infancy and when the guarded filtration procedure is used in older children.[29] Close ophthalmologic evaluation from birth of SWS patients is good medicine.[30]

Emotional support for each SWS patient is important. We recommend patients and their families contact the helpful and informative Sturge-Weber Foundation, P.O. Box 418, Mt. Freedom, New Jersey 07970-0418, phone: 800-627-5482, email: webmaster@sturge-weber.com, or visit its website: www.sturge-weber.com.

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