

# Cancer Prone Disease Section

## Mini Review

## Sturge Weber syndrome

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### Identity

**Alias:** Encephalotrigeminal angiomatosis; Angio-encephalo-cutaneous syndrome

**Inheritance:** Sporadic, equal frequency in the sexes.

### Clinics

#### Phenotype and clinics

Sturge Weber Syndrome is a congenital condition characterized by an intracranial vascular anomaly, leptomeningeal angiomatosis, most often involving the occipital and parietal lobes. The anomaly can affect both cerebral hemispheres.

An ipsilateral facial cutaneous capillary vascular malformation usually affects the upper face in the V1 distribution of the trigeminal nerve.

Other findings include glaucoma, buphthalmos, enlargement of the choroid plexus and seizures.

Hemiparesis, hemiatrophy, hemianopia and strokelike events may occur contralateral to the cortical abnormality.

Venous stasis results in ischemia underlying the leptomeningeal angiomatosis leading to calcification and laminar cortical necrosis.

#### Neoplastic risk

For any infant with a facial port-wine stain, the risk of Sturge-Weber syndrome with associated angiomatosis of the leptomeninges or vessels of the eye is approximately 10%. The risk increases to 25% when the entire side of the face is involved and 33% when both sides of the face are affected by port-wine stain.

Bony and soft tissue hypertrophy can develop with structures underlying the port-wine stain.

Reports exist of tumors found in association with Sturge-Weber syndrome; however it is unclear whether Sturge-Weber syndrome is associated with an increased risk of other neoplasms.

#### Treatment

Treatment is symptomatic and focuses on seizure control with antiepileptics or surgery, symptomatic and prophylactic migraine management, glaucoma treatment to reduce intraocular pressure and laser therapy for facial cutaneous vascular malformation.

Anecdotal aspirin use is used when children have strokelike episodes.

#### Evolution

75-90% of children with SWS develop partial seizures by 3 years of age.

50-75% of children have developmental delay or mental retardation.

Possible complications include status epilepticus, prolonged strokelike episodes, hearing disorder, intractable headaches.

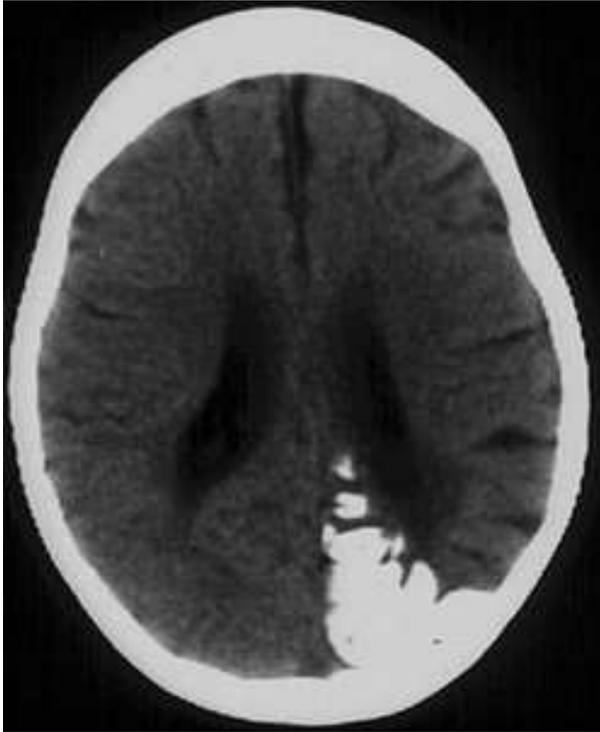
Glaucoma develops in 30-70% of individuals, and this along with visual field cuts can result in vision loss.

Neurologic deterioration in SWS is likely secondary to impaired blood flow to the brain and is worsened by seizures.

#### Prognosis

The prognosis for SWS patients is highly variable. Some children are minimally affected, if at all. Others have early onset seizures, numerous strokelike episodes and neurologic deterioration with hemiparesis and mental retardation.

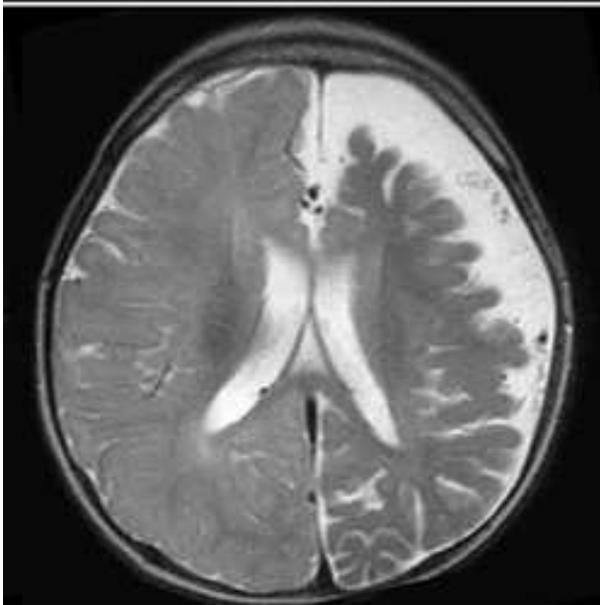
Life expectancy is thought to be normal.



An axial computed tomographic image revealing abnormal calcifications of the left occipital lobe in a 9 year old male patient.



A post gadolinium-enhanced axial T1 weighted image demonstrating prominent leptomeningeal enhancement over the right occipital lobe of a 7 month old male patient. The choroid plexus is enlarged in the posterior horn of the right lateral ventricle.



An axial T2 weighted MRI demonstrating left lateral frontal lobe atrophy in an 11 month old male patient.

Skull x-ray film may show classic "tram-line" or "tram-track" calcifications.  
 CT scan may show brain atrophy, calcification and ipsilateral choroid plexus enlargement.  
 MRI with gadolinium enhancement shows leptomeningeal angioma.  
 SPECT demonstrates decreased cortical perfusion.  
 PET demonstrates hypometabolism in areas that correspond to decreased perfusion.  
 EEG show electromagnetic changes in areas corresponding to the leptomeningeal angiomatosis.

## Cytogenetics

### Note

No consistent cytogenetic abnormalities have been found in association with Sturge-Weber syndrome.

One study found chromosomal abnormalities in 2 cultures derived from affected tissue compared to cultures from unaffected tissue of the same 2 individuals suggesting the presence of somatic mutation or chromosomal instability.

## Other findings

### Note

Microarray and reverse transcriptase-polymerase chain reaction data found an increase in fibronectin expression in fibroblasts derived from the port-wine stains of SWS subjects compared with fibroblasts from the normal skin of the same subjects. This finding suggests the presence of a somatic mutation affecting the port wine stain tissue.

## Genes involved and proteins

### Mutations

Somatic mutation has been suggested by Happle and others to have a role in Sturge-Weber syndrome because of its sporadic occurrence and unilateral localized distribution.

No prenatal or environmental risk factors have been identified.

A putative mutation has not yet been identified.

## References

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