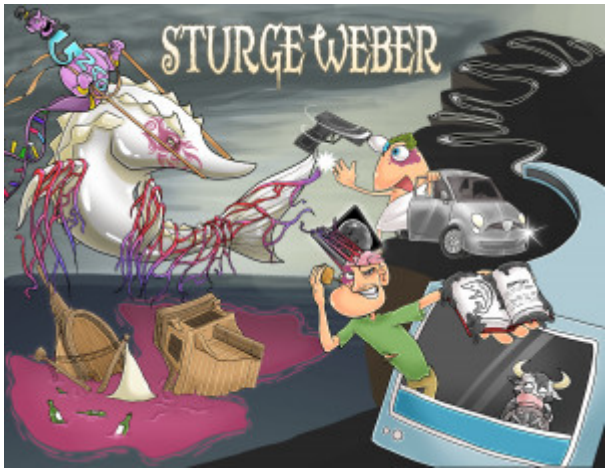
Sturge Weber Syndrome

Sturge-Weber syndrome, sometimes called encephalotrigeminal angiomatosis, is a rare congenital neurocutaneous disorder characterized by proliferation of arteries in the brain, resulting in multiple angiomas. Specifically, patients demonstrate leptomeningeal angiomas, along with arteriovenous malformations (AVMs). This syndrome is often associated with port-wine stains on the face, ipsilateral to the leptomeningeal angioma, along with neurologic consequences, like seizures, varying degrees of mental retardation, and glaucoma. Normally, all manifestations of this disease are on the same side of the head. There is also an increased incidence of pheochromocytomas. On CT, patients with Sturge-Weber syndrome display tram-track calcifications, which indicate cortical and subcortical calcifications.



PLAY PICMONIC

Pathophysiology

GNAQ Gene

G-neck Genie

Sturge-Weber syndrome is not inherited, but rather occurs sporadically as a result of a somatic activating mutation occurring in the GNAQ gene.

Signs and Symptoms

Port Wine Stain

Port red Wine

Port-wine stains, also called nevus flammeus, or firemarks, are birthmarks that occur as a result of capillary malformations in the skin. Port-wine stains tend to grow proportional to the growth of a child, rather than fading. They may present as a red color, maturing to a dark red, or purple color with age. These can present anywhere on the body, but are most often seen on the face. Lesions in the trigeminal nerve distribution are sometimes associated with Sturge-Weber syndrome.

Arterial Venous Malformation

Web of Arteries and Veins

In Sturge-Weber syndrome patients are subject to various permutations of arteriovenous malformations (AVM). These include capillary malformations, leading to facial port-wine stains, along with malformations in the retinal vessels, pia mater and choroid in the area of the trigeminal nerve.

Leptomeningeal Angiomas

Laptop in Meninges

Patients with Sturge-Weber syndrome may develop abnormal proliferation of blood vessels in the leptomeninges, which refer to the pia mater and arachnoid mater of the brain and spinal cord. Commonly, patients with Sturge-Weber syndrome have leptomeningeal angiomas on the ipsilateral side as their port-wine stain.

Intellectual Disability

Tar Covered Book

In Sturge-Weber syndrome, neurological concerns relate to the extent of angioma growth on brain tissue. This may result in neurologic consequences, such as developmental delay, or intellectual disability.

Seizures

Caesar

In this syndrome, angiomas (occurring as a result of arteriovenous malformations) create abnormal conditions for brain function. As a result, patients with Sturge-Weber syndrome may display seizure activity, and this is the most common early neurologic manifestation, often starting by one year of age. Patients may have initial focal seizures progressing to frequent, secondary generalized seizures, along with increasing seizure frequency and duration, despite the use of antiepileptic drugs. Therapies include surgical intervention, vagal nerve stimulation and anticonvulsant drug therapy.

Pheochromocytoma

Phiat-chrome

Pheochromocytomas are adrenal gland tumors that secrete catecholamines. Patients with Sturge-Weber syndrome have a higher predisposition towards developing pheochromocytomas than the normal population.

Glaucoma

Glock-eye

Those with Sturge-Weber syndrome often have ocular involvement, typically in the form of glaucoma, a disorder of increased fluid pressure in the eye. These most commonly present ipsilaterally to the site of the patient's port-wine stain, and patients can display vision loss, strabismus, myopia, visual field defect, optic nerve cupping and corneal edema.

Tram-Track Calcifications

Tram-Track

On CT scan, patients with Sturge-Weber syndrome display tram-track calcifications, which indicate cortical and subcortical calcifications.