

Sturge-Weber Syndrome

Sturge-Weber Syndrome (SWS) is a rare congenital neurocutaneous disorder characterized by abnormal blood vessel development in the skin, eyes, and brain. This syndrome is associated with genetic mutations, specifically in the GNAQ gene, which results in the abnormal growth and dilation of blood vessels, known as angiomas. The manifestations of SWS can vary significantly in severity, with clinical features ranging from isolated port-wine stains to severe neurological impairments. Early diagnosis and multidisciplinary management are essential for optimizing outcomes.

Etiology and Pathophysiology

SWS is caused by a somatic mutation in the GNAQ gene, which leads to the abnormal development of blood vessels in the affected areas. The mutation results in the formation of angiomas—dilated blood vessels that can affect the brain, skin, and eyes. While the precise mechanisms underlying the vascular malformations are still under investigation, these angiomas are believed to disrupt normal blood flow and increase pressure on surrounding tissues, leading to a variety of clinical complications.

- **Skin Involvement:** The hallmark feature of SWS is the presence of a port-wine stain (PWS), a vascular malformation that appears as a red to purple discoloration of the skin, typically affecting one side of the face. The PWS often follows the distribution of the trigeminal nerve, involving the forehead, eyelid, and cheek. Although this birthmark may remain static in size or slightly grow in the early years of life, it may darken, thicken, or elevate over time. Bilateral port-wine stains are rare but may occur in some cases of SWS.
- **Neurological Involvement:** The vascular malformations in SWS extend to the central nervous system (CNS), where they are most commonly localized to the cerebral cortex and meninges. These angiomas can cause a range of neurological complications, including seizures, hemiparesis (paralysis on one side of the body), developmental delays, and cognitive impairments. The pressure exerted by the angiomas on the brain tissue can disrupt normal neurological function and contribute to long-term morbidity.
- **Ophthalmic Involvement:** The increased blood flow to the ocular region can lead to glaucoma, a condition characterized by elevated intraocular pressure that damages the optic nerve, resulting in visual impairment and potential blindness if untreated. Ophthalmologic evaluation is critical in patients with SWS to monitor for the development of glaucoma.

Clinical Features and Diagnosis

The clinical diagnosis of SWS is primarily based on the presence of a port-wine stain and associated neurological or ophthalmic manifestations. The skin changes typically present at birth or in early infancy, and the distribution of the port-wine stain is usually limited to one side of the face. In rare cases, bilateral involvement or involvement of other body regions may be observed.

Diagnosis can be confirmed through neuroimaging studies such as magnetic resonance imaging (MRI), which can identify cerebral angiomas and assess the extent of brain involvement. Angiography may also be performed to evaluate the vascular malformations more precisely. Genetic testing for GNAQ mutations may be helpful in confirming the diagnosis in some cases, though it is not universally recommended.

Multidisciplinary Management

The management of SWS involves a multidisciplinary approach, as the syndrome affects multiple organ systems. The following specialists typically play a role in the care of patients with Sturge-Weber Syndrome:

- **Neurologist:** A neurologist should be involved to assess and manage neurological symptoms, particularly seizures and developmental delays. Anticonvulsant medications such as levetiracetam and valproate are commonly prescribed to control seizure activity. In some cases, surgical interventions, such as resection of the vascular malformations, may be considered for refractory seizures or significant brain involvement.
- **Ophthalmologist:** Given the risk of glaucoma in patients with SWS, regular ophthalmologic examinations are essential. The ophthalmologist can monitor for elevated intraocular pressure and initiate treatment with eye drops such as prostaglandin analogs (e.g., latanoprost) to lower intraocular pressure and prevent further vision loss. In more severe cases, surgical interventions, including trabeculectomy or glaucoma drainage devices, may be required.
- **Dermatologist:** The port-wine stains in SWS can lead to cosmetic concerns and functional complications if they obstruct the eyes or other facial features. Treatment options for port-wine stains include laser therapy and, in severe cases, surgical excision.

Treatment Options

- **Laser Therapy:**
 - **Pulsed Dye Laser (PDL):** The pulsed dye laser is the primary treatment for port-wine stains. It works by emitting focused bursts of light that target the hemoglobin in the blood vessels, causing them to shrink and reducing the redness of the birthmark. Multiple treatments are often necessary, and the success of laser therapy depends on the size, location, and age of the port-wine stain.

- **Fractional CO2 Lasers:** In cases where the PDL is ineffective, fractional CO2 lasers may be considered for thicker or more elevated lesions. This laser type helps with tissue resurfacing and reduces the appearance of the port-wine stain by targeting deeper layers of the skin.
- **Surgical Intervention:** In cases where laser therapy is insufficient or where the vascular malformations cause significant functional impairment, surgical options such as tissue excision or reconstruction may be necessary. Cranial surgery may be performed to remove or alleviate pressure from the cerebral angiomas, particularly in patients with significant neurological involvement.
- **Seizure Management:** Pharmacologic treatment with anticonvulsants is essential to control seizures in patients with neurological involvement. Medications like levetiracetam and topiramate are frequently used. If seizures are refractory to medication, more invasive treatments such as vagus nerve stimulation (VNS) or epilepsy surgery may be considered.

Prognosis

The prognosis of Sturge-Weber Syndrome is highly variable and depends on the extent of the disease. Patients with mild forms of the syndrome, with limited port-wine stains and no significant neurological or ophthalmic involvement, may have a near-normal life expectancy and quality of life. However, individuals with extensive brain involvement, severe seizures, or glaucoma may experience significant neurological deficits and diminished quality of life. Early diagnosis and intervention are key to improving outcomes, particularly in managing seizures and preventing vision loss from glaucoma.

Conclusion

Sturge-Weber Syndrome is a rare and complex condition that requires a multidisciplinary treatment approach to manage its diverse clinical manifestations. Advances in laser therapy and seizure management have significantly improved the quality of life for many patients. However, ongoing research is needed to further understand the genetic and molecular mechanisms underlying SWS and to develop more effective treatments, particularly for the neurological complications of the syndrome.

References

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- ❖ Parker, W. G., Roth, J. L., & Morton, D. K. (2021). Surgical interventions in Sturge-Weber syndrome: Outcomes and considerations. *Journal of Neurosurgery Pediatrics*, 27(2), 132-140. <https://doi.org/10.3171/2021.2.PEDS20483>