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The SWSC/SWSCC is a branch of the Vascular Birthmarks Foundation

Sturge-Weber Syndrome Community

About Sturge-Weber Syndrome

Publication of the Sturge-Weber Syndrome Community and the Vascular Birthmarks Foundation

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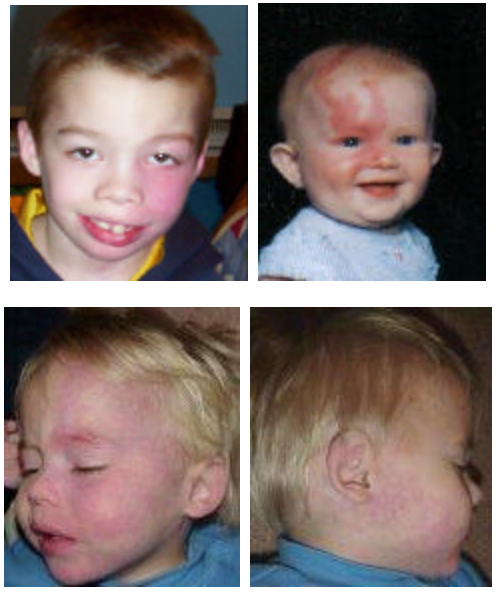
About Sturge-Weber Syndrome

Sturge-Weber Syndrome (**SWS**) is a congenital disease (present at birth). There is no present known cause or cure for the disease. Not all cases of **SWS** are identical and symptoms and their severity can vary.

A **Port Wine Stain** (**PWS**) on the face is usually visible at birth. The **PWS** most often covers the forehead and eyelid, but can include a larger portion (or smaller) on one or both sides of the face and head, and can extend into areas like the throat, nose, tongue, gums, ear canal, or extremities. However, the patterns and severity of **PWS** will vary on a case-to-case basis. In rare cases, there is no visible **PWS**.



The forehead and eye areas are the most common places to find a PWS associated with SWS, however, the patterns of PWS can vary greatly in each patient.

Notes

Port Wine Stain (PWS) – PWS is present at birth, and is a mass of malformed and dilated blood vessels in the skin.

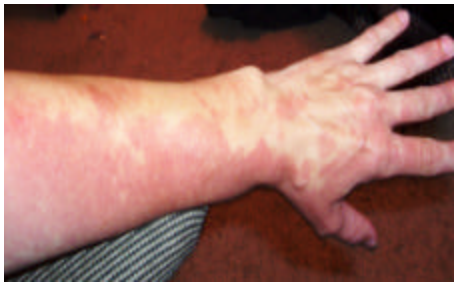
Retardation – Depending on severity, some of the symptoms might include the following: The limited ability to care for oneself, low IQ, or problems with communication and learning.

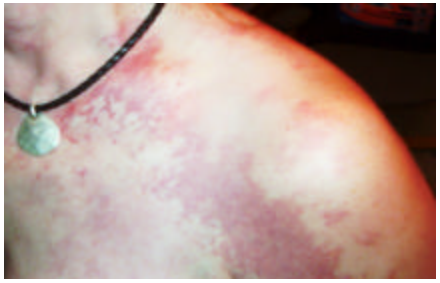
Seizures – There are many different types of seizures, and some patients can experience one or more of these types. The area of the brain affected can determine the type, severity and frequency of seizure activity. Patients are diagnosed on an individual basis. Signs of seizure can include, but aren't limited to, one or more of the following: loss of consciousness, staring, suddenly collapsing and falling, jerking or convulsing of the face or limbs, or the stiffening limbs. Consult a physician for information and diagnosis.

Sturge-Weber syndrome (SWS) – A disease present at birth (congenital), usually defined by a PWS on the face, and can include a variety of symptoms such as brain involvement, seizures, and glaucoma.

Unilateral – Affecting one side.

Vagus Nerve Stimulator (VNS) – A device implanted in the chest with two wires leading to the vagus nerve in the left side of the neck. A magnet is also used with the VNS. When a seizure is sensed to be coming on, the magnet is passed over the implanted device, generating extra stimulation to interrupt the seizure. This treatment is not considered a cure for seizures.





There are reported cases of **SWS** where no visible **PWS** is present, and others where **PWS** is also present on the trunk and/or extremities.

SWS is generally characterized by the presence of **leptomeningeal angiomas**, abnormal blood vessels on the outer layer of the brain. The presence of the **PWS** on the outer layer of the brain can result in **calcification** of the brain, and **atrophy** of the brain tissue.

Seizures, headaches and migraines are also a common symptom of SWS.

Most **seizure** activity can be controlled or modified by the use of medication. In the most severe cases, a **hemispherectomy** (where one hemisphere of the brain is removed or detached) is used as a "last ditch effort" to stop uncontrolled seizures.

A device known as the **Vagus Nerve Stimulator** may also be implanted. The device stimulates the vagus nerve in the neck, sending a signal to the brain to help interrupt seizure activity.

Hemiparesis (the weakening or loss of use on the side of the body opposite the **PWS**) can also be present.

In rare cases **SWS** can affect other organs in the body.

Mild to severe **retardation** can also be a result of **SWS**. **Learning disability** can be another factor in **SWS**, as well as behavioral problems, and in some cases, symptoms similar to **ADD/ADHD** can be present.

Glaucoma may be present at birth, or can appear months or years later. This disease affects the eye, resulting in vision loss due to damage to the optic nerve. It is recommended that patients have a yearly ophthalmology examination for **glaucoma**. Even **if glaucoma** is not present early on, close monitoring of the eyes for symptoms of this disease should continue throughout life.

SWS is typically a progressive disease. Some patients have been given the diagnosis of **SWS** when they only have **glaucoma** and a facial **PWS**, but brain involvement is typically the defining factor for **SWS**.

With PWS on the eyelid or near the eye, glaucoma can appear at any age. Patients should have a yearly eye exam to check for signs of glaucoma.

Choroidal hemangioma, located in the blood vessel layer beneath the retina (choroid), is also related to SWS; as well as **buphthalmos**, an enlarged eye seen in infantile glaucoma.

De-bulking – The process of surgically removing excess tissue from areas like the lip.

EEG (electroencephalogram) – Test used to detect abnormal electrical activity of the brain.

Glaucoma – Eye disease, which causes increased fluid pressure and damage to the optic nerve. There are often no symptoms and vision loss can be rapid.

Hemiparesis – The weakness or slight paralysis of one side of the body.

Hemispherectomy – The removal of one hemisphere or lobe of the brain.

Laser (**Light Amplification by Stimulated Emission of Radiation) –** Treatment for PWS by use of the transmission of an intense beam of bright light to the affected area. There are many types of lasers and laser manufacturers. Consult your physician for more information.

Learning Disability – Including, but not limited to, the difficulty with reading, spelling, language, math, etc.

Leptomeningeal Angiomas – Mass of abnormal blood vessels on the outer layer of the brain (meninges).

Magnetic Resonance Imaging (MRI) – The generation of a highly detailed 2-dimentional or 3-dimentional image of tissues inside the body using a magnetic field.

Nodules – A knoblike growth or bump, protruding from the skin.

Definition of Terms Used

Attention Deficit Disorder/Attention Deficit Hyperactivity Disorder (ADD/ADHD) – Disorder of the central nervous system. Patients can have trouble concentrating, paying attention, and processing information and stimulation. Children often exhibit problems in school, with the inability to sit still or follow directions, and with speaking out at inappropriate times.

Atrophy – The deterioration or shrinking of tissue.

Bilateral – Affecting both sides.

Blebs – A blister or pustule.

Buphthalmos - Enlarged eye (seen with infantile glaucoma)

Calcification – The hardening of tissue (brain tissue in relation to calcification regarding SWS).

Choroidal Hemangioma – Growth located in the blood vessel layer beneath the retina (choroid).

Computed Axial Tomography (CAT Scan) – The creation of a computer generated cross-sectional image from an X-ray.

Cobblestones – Blebs and nodules in PWS are often referred to as cobblestones. Bumps under the skin can look like small pebbles or cobblestones, creating an uneven skin texture.

Early treatment of the **PWS** is recommended, as the **PWS** can thicken over the years and develop **nodules** or **blebs** (sometimes referred to as **cobblestones**), which can bust open and bleed.

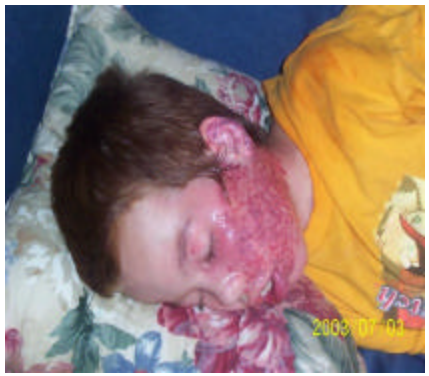
Laser treatments are now begun on infants, and can greatly improve the appearance and reduce affects of the **PWS** in years to come.

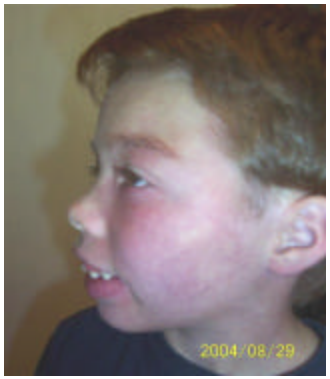
Also, early treatment of **PWS** of areas like the gums and lip are important, as these can become problem areas as they become engorged and grow larger. Affected gums can result in dental problems (such as bleeding of the gums, tooth decay and gum overlapping of teeth), and the lip area may require surgery for **de-bulking**.

The development of **laser** treatment has been greatly advanced over the years, and it is recommended that adults, who have never been treated or had little treatment, seek the advice of a skilled **laser** surgeon.

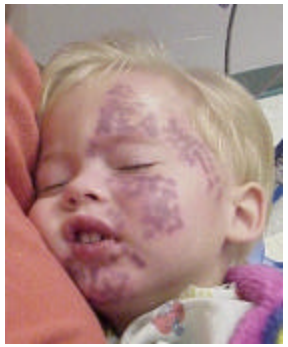
Many adults have received little or no treatment for port wine stain issues. As one ages, nodules or blebs can form, which bust and bleed. The skin can also thicken and darken. It is recommended that adults seek the advice of a physician.

Laser treatments usually leave small round temporary spots on the treated area, ranging in color from red to purple/black. There may also be swelling in the treated area. These symptoms usually begin to subside within several days, and it may take weeks or months for the full affects of **laser** treatment to be seen. In some cases, the **laser** doesn't leave any visible "dots" and no swelling is present. These are usually very superficial treatments.





Each port wine stain treatment, results, and effects depend on many factors - including location, depth of **PWS**, type of **laser** and settings, etc.



Laser treatment results can vary from patient to patient. Factors affecting the outcome range from type of laser used to the location and depth of the port wine stain on the face, trunk or extremities.

Some port wine stain syndromes and conditions can appear to be similar, and can be difficult to diagnose.

Magnetic Resonance Imaging (MRI) studies are typically used in the diagnosis of **SWS**. An **MRI** with contrast (using dye) is the preferred method in determining **SWS**. In addition, a **CAT Scan**, **EEG**, and other tests may be necessary in the treatment and diagnosis of **SWS** and its related conditions.

Resources for Sturge-Weber Syndrome

- **Sturge-Weber Syndrome Community**
  P.O. Box 24890
  Lexington, KY 40524-4890
  (859) 272-3857
  http://swscommunity.org

- **Sturge-Weber Syndrome Community Canada**
  Providing information in French and English languages.
  P.O. Box 79177
  Gatineau, QC.
  J8Y - 6V2
  http://swscommunitycanada.org

- **Sturge-Weber Syndrome Community Support Group MSN Groups – Public**
  http://groups.msn.com/SturgeWeberSyndromeCommunitySupport

- **Sturge-Weber Syndrome Community Support Group Yahoo Groups – Private – (SWC Talk Community)**
  http://health.groups.yahoo.com/group/swctalkcommunity/

- **Vascular Birthmarks Foundation**
  P.O. Box 106
  Latham, NY 12110
  (877) 823-4646
  http://birthmark.org