STURGE-WEBER SYNDROME FACT SHEET

Sturge-Weber syndrome (SWS) is a rare neurological and skin disorder characterized by nervous system problems and a permanent birthmark, known as a Port Wine Birthmark (PWB), usually on the face.¹ SWS affects approximately 1 in 40,000 to 1 in 400,000 globally² and fewer than 200,000 in the U.S.³

WHAT CAUSES STURGE-WEBER SYNDROME?

SWS is caused by a mutation of the GNAQ gene that occurs after conception and is not inherited. PWBs are caused by abnormally dilated capillaries in the skin, which produce red to purple discoloration.⁴

COMPLICATIONS ASSOCIATED WITH STURGE-WEBER SYNDROME MAY INCLUDE:

<table>
<thead>
<tr>
<th>Seizures and Convulsions⁵</th>
<th>Seizures and convulsions occur in 72%-80% of SWS patients with lesions on one side of the brain and 93% of patients with lesions on both sides of the brain. Seizures can begin anytime from birth to adulthood: ● 75% within the first year of life, 86% by age 2, 95% before age 5</th>
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<tr>
<td>Majority of Sturge-Weber patients are born with a Port Wine Birthmark⁵</td>
<td>A person born with a PWB has an 8%-15% chance of having SWS. The risk of SWS increases to 25% in patients with a birthmark covering half of the face.</td>
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<td>Glaucoma⁶</td>
<td>About 50% of patients with SWS will develop glaucoma if the birthmark involves the trigeminal area of the eye.</td>
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<td>Migraines and headaches⁷</td>
<td>44% to 62% of patients suffer migraines and headaches.</td>
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<tr>
<td>Developmental Delays⁷</td>
<td>50% to 75% of patients will have development delays.</td>
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<td>Mood/behavior problems⁸</td>
<td>Present in 85% of those with seizures and 58% without.</td>
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<tr>
<td>Paralysis⁷</td>
<td>Approximately 25%-56% of people with SWS have weakness or paralysis on the opposite side of the Port Wine Birthmark.</td>
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<td>Forme Fruste⁹</td>
<td>A complication of Type 3 SWS characterized by vascular malformation in the brain, with no facial birthmarks and usually no development of glaucoma. Forme Fruste is identified through brain scans.</td>
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<tr>
<td>Hormonal Abnormalities¹⁰</td>
<td>Patients with SWS may also experience hormonal abnormalities, which can increase the risk of hypothalamic–pituitary dysfunction.</td>
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WHO IS AT RISK FOR STURGE-WEBER SYNDROME?

SWS affects all races and sexes equally, with no clear genetic pattern. Two cases of SWS almost never arise in the same family.⁵ One in 50,000 people born in the U.S. will have SWS.⁷,⁸

HOW IS STURGE-WEBER SYNDROME DIAGNOSED?

SWS is usually diagnosed with a presence of a PWB on the upper eyelid or the forehead combined with glaucoma, abnormal blood vessels in the brain, or both. SWS can manifest at any time. Children with PWB accompanied by neurological signs/symptoms should receive a neurological evaluation. Additional signs of SWS may include:⁶

● Seizures, early handedness or evidence of a visual gaze preference by age two
● Neurological symptoms can start in later childhood or even in adulthood
● Glaucoma can begin at any time; at-risk individuals should be examined by an ophthalmologist every three months for the first few years and at least annually for life
HOW IS STURGE-WEBER SYNDROME TREATED?

Treatment of SWS is based on one's symptoms and associated underlying conditions:\(^1\)

- **Seizures**
  - Anticonvulsant medications to control and combat seizures
  - Neurosurgery to remove or disconnect the affected part of the brain when anticonvulsant medications do not prove effective
- **Port Wine Birthmark**
  - Laser therapy used to lighten or diminish the Port Wine Birthmark
- **Glaucma**
  - Eye drops are used to treat less severe cases
  - Surgery may be performed on patients with more serious cases of glaucoma
- **Educational therapy** prescribed for those with developmental delays
- **Physical therapy** for paralysis or weakness

WHAT RESEARCH IS BEING CONDUCTED INTO STURGE-WEBER SYNDROME?

Since its inception, the Sturge-Weber Foundation has been instrumental in researching and developing therapies to control the complications of Sturge-Weber syndrome. In May 2013, the Brain Vascular Malformation Consortium discovered the GNAQ gene mutation as a cause of SWS.\(^4\) Current research builds upon past research and focuses on validating new screening tools, evaluating new treatment strategies and efforts to determine the cause of SWS.

HOW CAN I GET INVOLVED AND FIND OUT MORE INFORMATION ABOUT STURGE-WEBER SYNDROME?

Since established in 1987, the Foundation has been the leading non-profit seed grant funder, making huge strides in stimulating interest in research for SWS and other vascular diseases. The Foundation continually seeks new breakthroughs in preventing medical and developmental problems resulting from the condition. People can help in many ways including:

- **Volunteer**
- **Donate tissue samples**
- **Donate funds**
- **Hold an awareness event**
- **Attend an educational forum**
- **Participate in research**

For more information about becoming involved, visit [www.sturge-weber.org/get-involved/general-info/get-involved.html](http://www.sturge-weber.org/get-involved/general-info/get-involved.html).

For more information on SWS, education programs and to access the online Learning Center, visit [www.sturge-weber.org/learning-center.html](http://www.sturge-weber.org/learning-center.html).

ABOUT THE STURGE-WEBER FOUNDATION

The Sturge-Weber Foundation’s international mission is to improve the quality of life and care for people with Sturge-Weber syndrome and associated port-wine birthmark conditions through collaborative education, advocacy, research and friendly support.

REFERENCES


