The Sturge-Weber Foundation

THE MISSION
What Causes SWS?
SWS is caused by a mutation of the GNAQ gene that occurs after conception. It is not inherited. PWBs are caused by abnormal dilated capillaries in the skin, which produce red to purple discoloration.

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

How is SWS Diagnosed?
SWS is usually diagnosed with a presence of a PWB on the upper eyelid or the forehead accompanied by neurological signs/symptoms manifest at any time. Children with PWB development, only 8% develop SWS. 85% of children with SWS have PWB covering half of the face.

Development Delay: 50-75%
Children With SWS: 72-93%
Complications Associated With SWS:
- Seizures/convulsions can begin anywhere from birth to adulthood. 75% within the first year of life; 85% by the age of 2, 95% before age 5.
- Majority of SWS patients are born with a PWB. The risk of SWS increases to 25% in patients with a PWB covering half of the face.
- Glaucoma will develop in 50% of patients if the PWB involves the trigeminal area of the eye.
- Migraines and headaches are common among SWS patients and can be severe. Development delays occur in 50-75% of SWS patients.
- Mood/behavior problems are present in 85% of patients with seizures and 58% of the face.
- Weakness or paralysis occurs in 25-56% of SWS patients on the opposite side of the PWB.
- Weakness or paralysis occurs in 25-56% of SWS patients on the opposite side of the PWB.
- Forme Fruste occurs in 25-56% of SWS patients on the opposite side of the PWB.
- Seizures/convulsions can begin anywhere from birth to adulthood. 75% within the first year of life; 85% by the age of 2, 95% before age 5.
- Seizures/convulsions can begin anywhere from birth to adulthood. 75% within the first year of life; 85% by the age of 2, 95% before age 5.

Experience Headaches: 44-62%
Develop Glaucoma: 30-71%
PWB is more common than SWS. Of the 15% of people with PWB development, only 8% develop SWS. 85% of SWS cases only affect one side of the body or brain.

Additional signs of SWS may include:
- Seizures, early handedness or evidence of a visual 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 scans.

SWS FACTS AND STATISTICS

The SWF Mission: Since 1987, The Sturge-Weber Foundation’s international mission is to improve the quality of life and care for people with SWS and PWB conditions through collaborative education, advocacy, research and friendly support.

The SWF Research Initiative
SWS has been instrumental in researching and developing therapies to control the complications of SWS in the areas of dermatology, ophthalmology, and neurology. SWF strives to stimulate and support research on all aspects of SWS. Based on these efforts, we have identified a clear set of research priorities to enable major breakthroughs in understanding the biological basis of SWS, as well as the first tests of therapeutic options.

SWF Accomplishes This Through:
- In 2013, the Brain Vascular Malformation Consortium (BVMC) discovered the GNAQ gene mutation as a cause of SWS. SWF was instrumental in this discovery through its continuous support in researching and developing therapies to control the complications of SWS.
- Research Grants relating to epigenetics, gene expression, proteomics, neurobehavioral studies, and glaucoma have received funding as a result of research performed by SWS patients.
- SWF is proud to provide collaborative access to the SWF Ocular Surgery database and to the SWF Registry.

The SWF Clinical Care Network evolved out of the Foundation’s expanding network of healthcare professionals and scientists interested in improving the quality of life and care for patients with SWS and PWB. Each center is staffed by a team of specialists who coordinate in the evaluation and management of each patient.

The SWF Clinical Care Network also works on other diseases with vascular malformations to share information and basic research. This network model has been groundbreaking for rare diseases and the Foundation is seeing evidence that it will continue to have an impact on the research and development of an overall understanding of diseases with vascular malformations.

The SWF has 28 centers across the U.S. and abroad in the Clinical Care Network.

There is no cure for SWS.

Approved treatments include:
- Laser Surgery
- Anticonvulsant/Sleeping/Physical Therapy
- Glaucoma Medication
- Ocular Surgery
- Occupational/Speech/Physical Therapy
- Educational Therapy

THE SWF MISSION

THE SWF MISSION

THE SWF MISSION
SWF Education Programs and Support
Education and personal support are the Foundation’s main vehicles for increasing the understanding of SWS and PWB.
• SWF has a comprehensive, reader friendly website with relative subjects and resource links
• One on one friendly support to individuals, family members and care givers
• Physician referral at a convenient location
• Information about the importance of tissue donation
• Two Online Support Systems where participants share their stories, ask questions and provide support and comfort to one another
• Family days, education days, regional and international conferences offer opportunities to meet others and learn more about their mutual concerns from medical professionals.
• Volunteer and fundraising events that help spread awareness for the disease and contribute crucial funds to benefit the Foundation’s programs.

SWF Patient Advocacy
Advocacy is a main avenue for SWF to raise awareness. Through these efforts, SWF is able to help the public see past the disability to the person and enable families and individuals to obtain the medical care, employment, education, respect and personal achievement they seek. The Foundation serves as a conduit for global collaborations through numerous coalitions and umbrella organizations including the FDA, National Institutes of Health, American Brain Coalition and more.

To create a lasting impact, become a CHAMPION today!
Visit the SWF at www.sturge-weber.org or call 973.895.4445.