The Sturge-Weber Foundation

THE MISSION

Sturge-Weber Syndrome:

\ ‘sterj - ‘web-er- \ (SWS) - know as a rare neurological skin disorder characterized by nervous system problems and a permanent birthmark, knows as a Port Wine Birthmark (PWB), usually on the face. SWS affects approximately 1 in 40,000 to 1 in 400,000 globally and fewer than 200,000 in the U.S.

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 abnormally 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 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:

- 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 the first few years and at least annually for life.

Development Delay: 50-75%

Children With SWS Having Seizures: 72-93%

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.

Complications Associated With SWS

- **Seizures/convulsions** can begin anytime from birth to adulthood. 75% within the first year of life; 86% 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**[6] 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% without.
- **Weakness or paralysis** occurs in 25-56% of SWS patients on the opposite side of the PWB.
- **Forme Fruste** is a complication of Type 3 SWS characterized by vascular malformation in the brain with no PWB and usually no development of glaucoma. This condition is identified through brain scans.
- **Hormonal abnormalities** may also be experienced which can increase the risk of hypothalamic-pituitary dysfunction.

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[8].

THE STURGE-WEBER FOUNDATION RESEARCH NETWORK

The SWF Research Initiative

SWF has been instrumental in researching and developing therapies to control the complications of SWF 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 May 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 - gene expression of SWS, neuropsychiatric and behavioral issues, neurorehabilitation, and glaucoma and blood flow studies targeting the adult SWS population.
- Provides collaboration of data among researchers in all associated SWS medical fields.

THE STURGE-WEBER FOUNDATION CLINICAL CARE NETWORK

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 living with SWS and PWB. Each center is staffed by a team of specialists who collaborate 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.

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SWF has 28 centers across the U.S. and abroad in the Clinical Care Network and it is still expanding.

There is no cure for SWS.

Approved treatments include:

Laser Surgery
Anticonvulsant Medications
Neurosurgery
Glaucoma Medication
Ocular Surgery
Occupational/Speech/Physical Therapy
Educational Therapy

The SWF International Registry

The Foundation has an online International Patient Registry for individuals diagnosed with SWS or PWB in the forehead and/or eye region. Through this registry, vital information about individual patient conditions, complications and current treatments will help to identify new effective treatments to provide a better quality of life for those with SWS and PWB. The Registry is completely confidential and no personal information is shared with researchers or medical professionals without the patients explicit permission.

For more information and to participate in the registry, visit https://swsregistry.patient-crossroads.org.

THE SWF MISSION

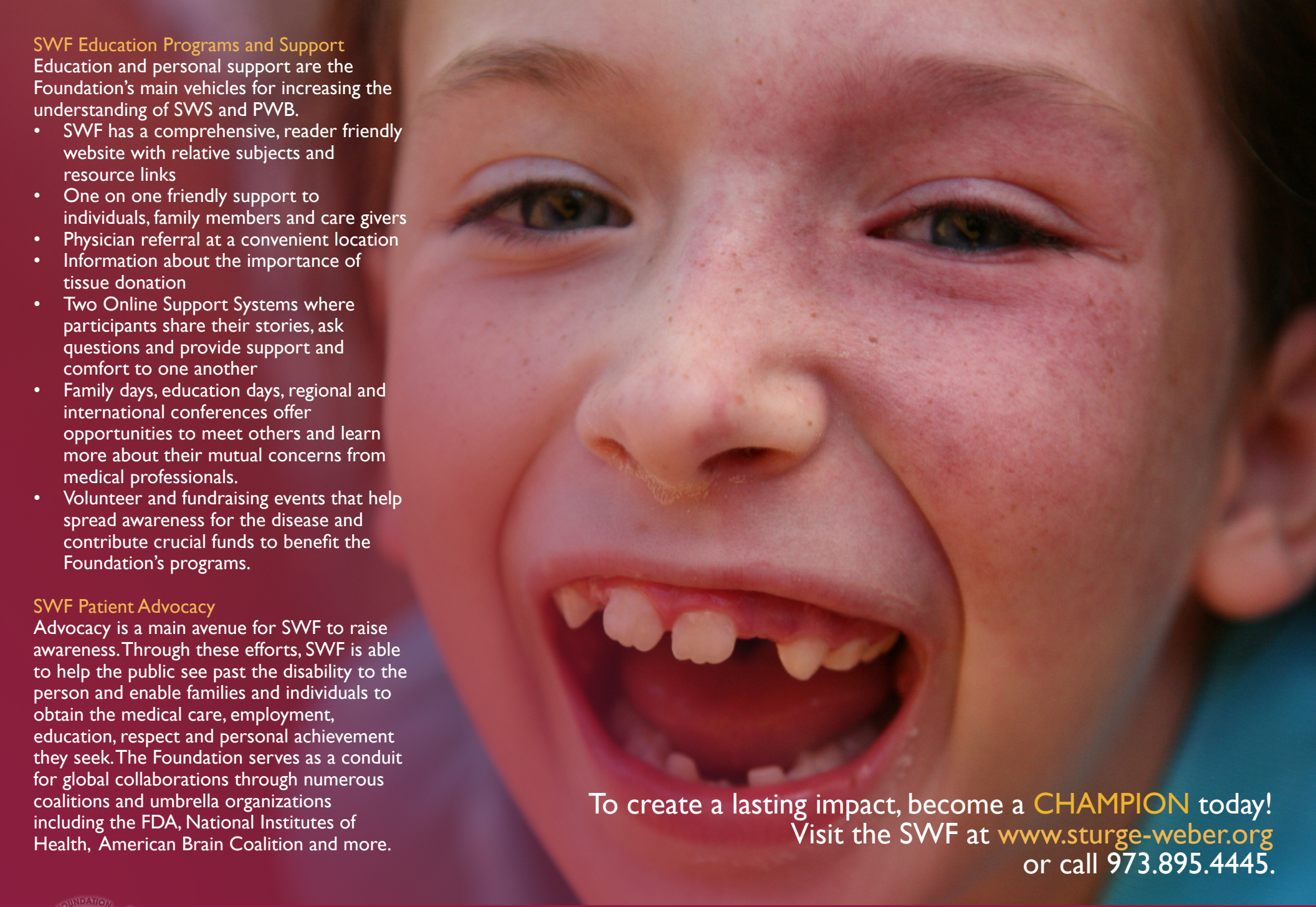
SWF Education Programs and Support

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.

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