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. SWS affects approximately 1 in 40,000 to 1 in 400,000 globally and fewer than 200,000 in the U.S.

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.

COMPLICATIONS ASSOCIATED WITH STURGE-WEBER SYNDROME MAY INCLUDE:

<table>
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<th>Complication</th>
<th>Description</th>
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| Seizures and Convulsions                          | 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 |
| Majority of Sturge-Weber patients are born with a Port Wine Birthmark | 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. |
| Glaucoma                                         | About 50% of patients with SWS will develop glaucoma if the birthmark involves the trigeminal area of the eye. |
| Migraines and headaches                           | 44% to 62% of patients suffer migraines and headaches. |
| Developmental Delays                              | 50% to 75% of patients will have development delays. |
| Mood/behavior problems                            | Present in 85% of those with seizures and 58% without. |
| Paralysis                                         | Approximately 25%-56% of people with SWS have weakness or paralysis on the opposite side of the Port Wine Birthmark. |
| Forme Fruste                                      | 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. |
| Hormonal Abnormalities                            | Patients with SWS may also experience hormonal abnormalities, which can increase the risk of hypothalamic–pituitary dysfunction. |

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. One in 50,000 people born in the U.S. will have SWS.

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:

- 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

- **Glaucoma**
  - 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

The Sturge-Weber Foundation (SWF) was founded in 1987 to improve the quality of life and care for people with Sturge-Weber syndrome (SWS) and Port Wine Birthmarks (PWB). The Foundation is focused on research, collaborative education and advocacy.

**RESEARCH: THE STURGE-WEBER RESEARCH INITIATIVE**

The SWF has been instrumental in researching and developing therapies to control the complications of Sturge-Weber syndrome. The Foundation 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.

**RESEARCH GRANTS**

To help advance research and treatments of SWS, the SWF awards research grants relating to:

- Studies of the gene expression of SWS, clinical issues relevant to individuals with SWS
- Angiogenesis, neurogenesis, neuroendocrinology, neuropsychiatric and behavior issues
- Glucose metabolism and cerebral blood flow
- Neurobiology studies that are innovative and pursue new avenues of investigation related to all aspects of Sturge-Weber and Port Wine Birthmarks
- Glaucoma and blood flow studies investigations targeting the adult SWS population

**SWS CENTERS OF EXCELLENCE**

The Centers of Excellence 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 PWBs. Each Center is staffed by a team of specialists who collaborate in the evaluation and management of each patient.

These Centers of Excellence also work on other diseases with vascular malformations to share information and basic research. This consortia model has been groundbreaking for rare diseases and the Foundation is seeing evidence that it will continue to have a big impact on the research and development of an overall understanding of diseases with vascular malformations.

**STURGE-WEBER INTERNATIONAL REGISTRY**

The Foundation launched the online Sturge-Weber International Patient Registry for individuals diagnosed with SWS or PWB in the forehead and/or eye region. The goal is to identify effective treatments to help those with SWS and PWB experience life to the fullest. For more information and to participate in the registry, visit [www.swsregistry.patientcrossroads.org](http://www.swsregistry.patientcrossroads.org).
EDUCATION PROGRAMS AND SUPPORT

Education is the Foundation’s main vehicle for increasing the understanding of SWS and PWBs. The SWF offers numerous educational tools to increase awareness of SWS including:

- Disease-focused SWF Posters and Flyers for display in schools, doctor’s offices, places of worship, churches, etc.
- The *Sturge-Weber Syndrome* textbook
- Information about the importance of tissue donation
- *Branching Out* periodic newsletter, booklets for schools, colleges and medical schools

To access these materials, visit the Foundation website at [www.sturge-weber.org/library/patient-resources.html](http://www.sturge-weber.org/library/patient-resources.html). To learn about how to donate tissue for SWS research, visit [www.sturge-weber.org/studies-and-science/tissue-donation.html](http://www.sturge-weber.org/studies-and-science/tissue-donation.html).

In addition, the SWF hosts a number of volunteer and fundraising events each year to help spread awareness for the disease. Most recently, they hosted the 2014 Danny Benefit, an annual fundraising event focused on raising money to benefit the Foundation’s programs. To learn about or participate in upcoming Foundation activities, visit [www.sturge-weber.org/get-involved/events/volunteer-happenings-sp-7386488.html](http://www.sturge-weber.org/get-involved/events/volunteer-happenings-sp-7386488.html).

PATIENT ADVOCACY

Patient advocacy is a main avenue for the Foundation to raise awareness of SWS and related rare diseases. Through these efforts, our goal is 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 Coalition of Skin Diseases, Brain Vascular Malformation Consortium, FDA, National Organization for Rare Disorders, National Institutes of Health and American Brain Coalition.

WHAT’S NEXT FOR THE FOUNDATION?

The Foundation has an online Learning Center, allowing people access to information on their syndrome and available treatments. The Foundation is also planning a global meeting for organizations dedicated to vascular malformations.

HOW YOU CAN HELP:

You can support The Sturge-Weber Foundation’s efforts in many ways, including

- Become a Support Volunteer or Family Day Coordinator
- Hold an Awareness Event
- Create a FirstGiving Page
- Become an Advocacy, Local Publicity or Medical Exhibit Booth Volunteer
- Participate in Research and the International Registry
- Donate

For more details on how to get involved and support The Sturge-Weber Foundation, visit [www.sturge-weber.org](http://www.sturge-weber.org).

THE STRONGER THE WIND               THE TOUGHER THE TREE
aHUS Meetups: Houston and Chicago
aHUS Meetups: Uniting the aHUS Family

Houston

- 20 attendees
- 11 families represented
- 9 caregivers and 2 adult patients
- 1 patient not currently on Soliris therapy
- Average travel distance: 275 miles

Survey Statistics
- 55% of the families found out about the Houston Meetup from OneSource
- 50% of who took the survey wanted to learn more from OneSource
- 88% of attendees prefer to receive information through e-mail and/or OneSource

Chicago

- 44 Attendees
- 21 Families represented
- 15 caregivers, 6 adult patients
- 5 patients not currently on Soliris therapy
- Average travel distance: 277 miles

Survey Statistics
- 30% of the families found out about the Chicago Meetup from OneSource
- Attendees expressed a variety of ways they would like to receive information
- 62% of attendees prefer to receive information through e-mail and/or OneSource, 31% by mail
Microsite Offers One-Stop-Shop for aHUS Meetup Information

- Agenda clearly outlines meeting goals
- Opportunity for participants to submit questions in advance
- Photos and bios offer opportunity for participants to relate to presenters
- Online registration allows patients to quickly and conveniently register for meeting
- Patient stories helped participants learn other families journeys with aHUS

Online registration allows patients to quickly and conveniently register for meeting.
aHUS Meetup Materials Prepare All Stakeholders

- Equipped physicians, OneSource, RAMS with draft notes, HTML and printed invites to engage participants
- Visual consistency with logo selected by aHUS community
Social Media Activities Key in Spreading the Word and Recruiting Participants

- 20 posts on social media and advocacy sites
- 5 placements in advocacy group newsletters
- 3 websites featured the Meetups on their events section and main page
- 100% of posts included link to aHUS Meetup microsite
- 1,100+ followers were exposed to tweets that mentioned the aHUS Meetups
aHUS Family Connects Through Experiences

- Captured and posted video testimonials to microsite to help future meeting recruitment
- Conducted surveys at each Meetup so aHUS community can help shape and plan future events

The family whose daughter is 19 and was only diagnosed 2 months ago, just kept telling me, "We were hoping you’d be here…we had so many questions!"...I know they left the meeting with renewed hope and feeling strong about telling their doctor not to discontinue drug! These Meetups are a gift we’ve all been waiting for.

Christina Davis
Adult with aHUS

I loved coming to the Meetup. I felt really alone before … I got to meet a lot of people who have insights on how to help me live my life better with the disease.

Cheryl Biermann
Co-founder, Foundation for Children with Atypical HUS

Being together COUNTS - nothing can replace that piece of the human experience. Joining together common experiences, sharing our issues …it’s invaluable and a precious, rare thing. This meeting refreshed me, informed and inspired me so very much.

Linda Burke
Founder, Foundation for Children with Atypical HUS social networking site

My son is an aHUS patient. It was nice to hear from the doctors who really understand the disease and being able to ask questions. I would recommend this Meetup to anyone, it was a great experience.

Luiz
Parent of a child with aHUS

The feedback near the end of the Meetups was very positive and leaves me confident these regional meetings are not just informational, but good for our souls too.

Cheryl Biermann
Co-founder, Foundation for Children with Atypical HUS

Barbara Farcher
Parent of an adult with aHUS

This meeting refreshed me, informed and inspired me so very much.

Linda Burke
Founder, Foundation for Children with Atypical HUS social networking site