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Branching Out

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ROOTS TO A CURE

Special Section

FALL 2016

THE STURGE-WEBER FOUNDATION
MAGAZINE

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Branching Out

Volume 30 Number 4 | DECEMBER 2016

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On the cover: Tynan, Emily and Silas Rockin' Towards a Cure

Port-Wine Birthmarks (PWB) on the skin are developmental abnormalities in blood vessel formation (capillary malformations) that are more extensive and darker than the pink capillary birthmarks often seen at the nape of a baby's neck.

Sturge-Weber syndrome (SWS) is a rare congenital condition usually consisting of a facial port wine birthmark, glaucoma, and seizures, (although not all of these symptoms may be exhibited).

Klippel-Trenaunay syndrome, or KT, occurs as the result of a congenital vascular malformation in an extremity, such as an arm, leg, or foot.

The SWF is a clearinghouse of information for Port Wine Birthmarks, Sturge-Weber syndrome, and Klippel-Trenaunay syndrome.

THE SWF POLICY STATEMENT:

In implementing the purpose of The Sturge-Weber foundation to improve the quality of life for individuals with SWS and their families, The Foundation will act as clearinghouse of information, provide emotional support, and facilitate research on PWB, SWS and KT.

As a clearinghouse of information, The Foundation will seek information regarding management and treatment techniques and suggestions concerning education and emotional support and will facilitate the dissemination of appropriate information.

If, in facilitating research on PWB, SWS and KT. The Foundation provides financial or other support to a particular research project, The Foundation will base its decision upon need, The Foundation's financial resources, and medical advice.

The SWF is a member of the National Organization for Rare Disorders (NORD), American Brain Coalition (ABC), The Coalition of Skin Diseases (CSD), and the Association for Research in Vision and Ophthalmology (ARVO).

by Karen L. Ball, SWF, President & CEO

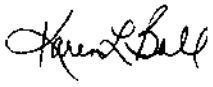
EXCITING CHANGES ARE COMING...

This issue brings you the most exciting updates on the SWF Champions who have been hard at work on your behalf. I can't think of a better gift than working with all the dedicated individuals around the world. We have made tremendous strides since I started SWF almost 30 years ago but as always I want it more and sooner as each precious life and challenge matter!

Check out the SWFIRN and PEN meeting summaries, the great Boston SWF Center of Excellence Educational Forum and all the tidbits that hold us together as we each take time to read, reflect and cherish our friends who make the SWF a global educational organization as well as a vital and welcome sanctuary.

We are eager to see you in Cincinnati this summer and wish you a blessed holiday of making cherished memories or reveling in times and loved ones from the past.

With faith, hope and love,



Karen L. Ball
President and CEO
The Sturge-Weber Foundation

OUR SWS PATIENT REGISTRY IS NOW UP! GO TO **WWW.SWSREGISTRY.ORG**
TO BE AWARE AND REGISTER YOURSELF OR YOUR CHILD. TOGETHER WE'LL MAKE SCIENTIFIC PROGRESS.

Centers of Excellence (COE)

These Centers provide the comprehensive care necessary for treating adults and children who have a port wine birthmark (PWB), Sturge-Weber syndrome (SWS) or Klippel-Trenaunay (KT).

Ann and Robert H Lurie Children's Hospital, Chicago, IL

Boston Children's Hospital, Boston, MA

Centro Medico de Puerto Rico, San Juan, PR

Cincinnati Children's Hospital Medical Center, Cincinnati, OH

Dell Children's Hospital, Austin, TX

Children's Hospital of Michigan, Detroit, MI

Mayo Clinic, Jacksonville, FL

Mayo Clinic, Phoenix, AZ

Mayo Clinic, Rochester, MN

NYU - Langone Medical Center, NY, NY

Nationwide Children's Hospital, Columbus, OH

Nemours Alfred I. duPont Hospital for Children, Wilmington, DE

Northeast Regional Epilepsy Group, Hackensack, NJ

Rady Children's Hospital, San Diego, CA

Texas Children's Hospital, Houston, TX

UNC Children's Hospital, Chapel Hill, NC

University of California, Irvine, CA

University of California, San Francisco, CA

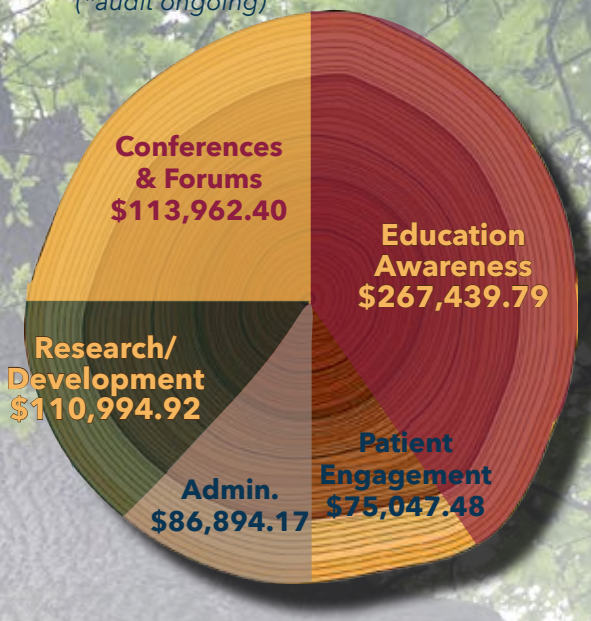
University of Illinois at Chicago Medical Center, Chicago, IL

University of Michigan - Mott Children's Hospital, Detroit, MI

Wills Eye Hospital, Philadelphia, PA

REVENUE: $600,113.66*

*(*audit ongoing)*

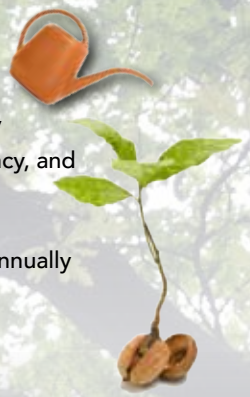


Patient Engagement Network (PEN)

Members and Staff who drive change!

Witney Arch
Kaelin Ball
Karen Ball
Michelle Daoust
Laura Embrey
Brian Fisher
Gloria Gomez
Mary Leonard
Jeffrey Needham
Madhurima Paturi
Candice Roberts
Kimberly Slater
Julia Terrell
Stephanie Tikkanen
Ann-Marie Vititoe

Continuous Cultivation - Roots to a Cure



The Sturge-Weber Foundation . . .

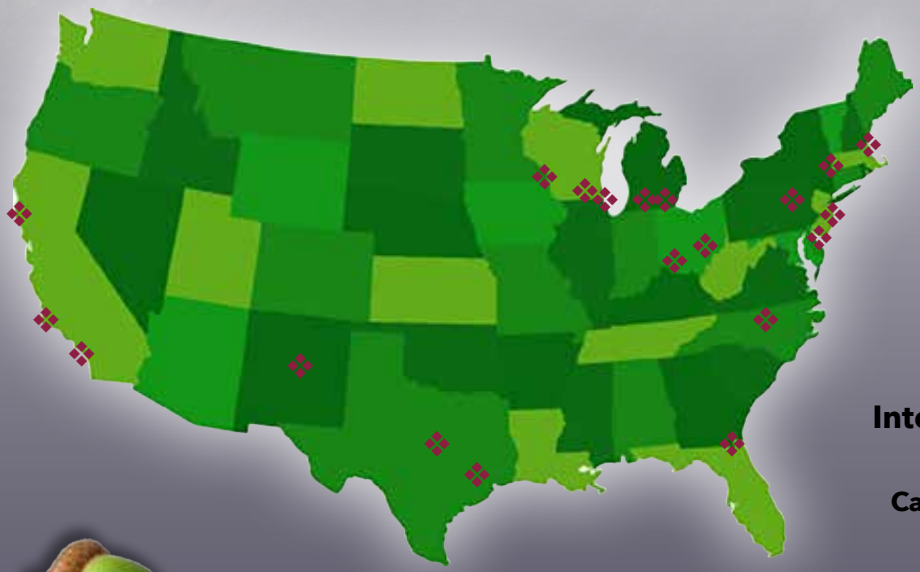
- attends **20** different society and organization meetings annually
- partners with **9** umbrella organizations with governance, advocacy, and specific intiatives
- interacts with **4** national institutes of health
- is involved in **2** consortiums or coalitions and attend meetings annually

3 out of 1,000

Only 3 out of 1,000 are born with a port wine birthmark.

3 to 9%

Only 3 to 9 % of people with a facial port wine stain are diagnosed with SWS.



462 out of 3921

There are only 3921 diagnosed SWS cases reported to SWF. Only 462 of those are registered on the **SWF International Registry.**

10 COUNTRIES **1 TERRITORY**

are represented in the **Sturge-Weber International Research Network** (SWFIRN).

This Year's Accomplishements

ROOTS TO A CURE

Special Section

FALL 2016

THE STURGE-WEBER FOUNDATION (SWF) IS GEARING UP TO CELEBRATE 30 YEARS OF SERVICE AND SUPPORT. THE SWF'S SUCCESS IN ACHIEVING GOALS ALONG THE WAY IS DIRECTLY DUE TO THE CARE, COLLABORATION AND CONNECTIONS WITH PATIENTS AND FAMILIES, VOLUNTEERS, HEALTHCARE PROVIDERS AND RESEARCHERS, FRIENDRAISERS IN LOCAL COMMUNITIES, VALUED CORPORATE SPONSORS AND THE NATIONAL INSTITUTES OF HEALTH. THIS ISSUE OF ROOTS TO A CURE HIGHLIGHTS THE PROGRESS TO DATE AND INFORMS THE READER OF THE EXCITING POSSIBILITIES FOR FUTURE DIRECTIONS. WE WELCOME YOUR SUGGESTIONS AND FEEDBACK AND AS ALWAYS ARE THANKFUL FOR YOUR FINANCIAL SUPPORT TO MAKE IT ALL POSSIBLE!

PATIENT CENTERED OUTCOMES RESEARCH INSTITUTE (PCORI)

The PCORI grant we received last February has been very instrumental in providing critical funding to gather leading patient advocates and researchers from around the world. The SWF Patient Engagement Network (PEN) is funded by the PCORI grant and the SWF convened patients and researchers in Atlanta, GA this past September to identify knowledge gaps, patient and family concerns, and set priorities to launch new strategic research initiatives.

THE STURGE-WEBER FOUNDATION
INTERNATIONAL RESEARCH NETWORK

SWF INTERNATIONAL RESEARCH NETWORK (SWFIRN)

The Sturge-Weber Foundation International Research Network (SWFIRN) and the SWF Patient Engagement Network (PEN) held a meeting in Atlanta from September 12-13, 2016. Co-chaired by **Doug Marchuk** (Duke University School of Medicine) and **Jonathan Pevsner** (Kennedy Krieger Institute; Chief Scientific Officer, SWF), and organized by **Brian Fisher** (VP of Operations & Corporate Partnerships, SWF), the meeting brought together a wide range of people committed to furthering research and care for people with Sturge-Weber syndrome. Here are some highlights of the SWFIRN research portion of the meeting.

We enjoyed presentations from three keynote speakers: **David Siderovski, J. Silvio Gutkind,** and **Veronica Kinsler.** All three are experts in G protein coupled receptors (GPCRs) and heterotrimeric G proteins including Gaq. Notably while all are experts in these areas, none have worked in the SWS field directly, and so we welcomed their perspectives on how to move the field forward.

- **What this means** (comments on the above paragraph for the non-scientist): *GNAQ* is one of 20,000 genes we have that make proteins (here the protein is called Gaq pronounced G alpha q). When any cell in your body receives information this often happens through a receptor which is a protein that sits on the cell surface. GCPRs are particular receptors that receive information and then send further messages into the cell to perform various tasks. We use receptors to respond to light, smells, adrenaline, transmitters, or hundreds of other substances. In the case of SWS, we know that Gaq is (1) connected to a handful of receptors that influence how cells grow and function, (2) connected to "signaling pathways" that help make an activated receptor transmit information downstream into the cell, (3) inappropriately turned on (activated), and (4) located in a particular type of cell, called an endothelial cell (as discovered by the groups of **Lan Huang** and **Joyce Bischoff,** as well as **Thuy Phung** who presented their findings here in Atlanta). Many researchers have studied Gaq in its own right, since its discovery in the 1990s. Other researchers have studied "G protein alpha subunits" of which we have 20—and Gaq is one of these 20. The other G protein alpha subunits are sometimes involved in other diseases. And the protein that is affected in SWS, Gaq, is mutated in a form of melanoma that many other researchers (outside the SWS community) study. We brought GPCR and Gaq experts who are not in the SWS field into this meeting as keynote speakers to help share their insights.

David Siderovski (West Virginia University, Health Sciences Center) provided a wonderful overview of GPCRs and G proteins, including the key family of "Regulators of G-protein signaling" (RGS) proteins that he discovered. He provided a broad context for our understanding of this field, and details of the role of Gaq in signaling. Topics ranged from a peptide inhibitor of G proteins to short interfering RNA to GTPase activing proteins (GAPs).

- **What this means** (comments on **David**'s topic for the non-scientist): David discovered a group of proteins (RGS) proteins that affect how Gaq works (and how other G protein alpha subunits work). We can think of Gaq as being one of dozens (or perhaps even hundreds) of interconnected proteins that help a cell grow and function. David explained the basic circuitry of how this works. We need to have this kind of understanding of what pathways are disrupted in SWS to know how to develop treatments. David discussed small compounds that might be used as drugs to help restore normal Gaq function, and he discussed other strategies to help fix the problems at the cellular level—therefore helping fix the symptoms of the person who has SWS.

J. Silvio Gutkind (UC San Diego, Moores Cancer Center), who originally identified GNAQ as an oncogene, discussed the role of GPCRs in cancer, including GNAQ in uveal melanoma. His inspiring talk emphasized the tremendous advances in our understanding of downstream signaling including potential targets such as MEK/ERK, PKC, TRIO, YAP, ROCK, FAK and PI3K. Both he and Catherine van Raammsdonk (who was unable to attend but was here is spirit) have developed mouse models of GNAQ that are relevant to uveal melanoma and may also be extended to SWS.

- **What this means** (comments on Silvio's topic for the non-scientist): In addition to making basic discoveries about *GNAQ* (such as the fact that it can mutate to become a cancer gene), Silvio showed that GPCRs can also mutate to become cancer genes. He went on to

show the details of how the pathways in these cells foster aberrant cell growth. His work has gone even further by leading to clinical trials—strategies to fight these disorders based on an understanding of the signaling mechanisms. The mutant mice that he and **Catherine van Raammsdonk** made (independently) are very important because animal models let us understand even more deeply how *GNAQ* mutations lead to changes at the levels of the cell and proteins and signaling pathways. Using animal models we can test drugs to determine if they can be helpful in patients with SWS.

Veronica Kinsler (UCL Great Ormond Street Institute of Child Health, London) provided an overview of the principles of mosaicism in disease. She described her remarkable studies of embryonic facial development, suggesting a new (and simpler) classification system to understand the facial distribution of port-wine birthmarks based on vascular embryological distribution rather than neural innervation of the face. At her clinic patients are seen with a broad range of vascular anomalies and her presentation included a variety of phenotypic associations in mosaic disorders. She noted mosaic activating mutations on *GNAQ* and *GNA11* in phakomatosis pigmentovascularis, and she modeled some of these mutations in a transgenic, mosaic zebrafish model.

- **What this means** (comments on **Veronica**'s topic for the non-scientist): "mosaicism" occurs when our body has the DNA (that includes genes) we inherited from our mother and father, and additionally we have DNA that has changed. Veronica described the basic principles of how mosaicism works. A basic question about port-wine birthmarks is whether their location predicts the occurrence of SWS (or how severe it will become), and her group introduced findings that the distribution of the birthmark follows the vasculature (rather than following the nerves). A location of a birthmark on the forehead is a predictor of an abnormal brain scan. Veronica also described mutations in GNAQ in other conditions related to SWS.

We enjoyed presentations from many other speakers. Very briefly:

Lan Huang (Boston Children's Hospital and Harvard Medical School) described an enrichment in GNAQ R183Q mutations in endothelial cells isolated from SWS brain specimens.

Gregory Tall (University of Michigan, Ann Arbor) discussed the role of GNAQ and GNA11 in ocular melanoma, and described folding of Gaq --and misfolding in the absence of Ric8A.

Brandon Farris (LobbyIt.com) discussed Capitol Hill updates and research funding.

Jonathan Pevsner and **Doug Marchuk** led a discussion of issues for the SWS research community (see the conclusions below).

Heather Etchevers (Aix-Marseille Université) discussed neurocutaneous malformations due to somatic mutations in the RAS-RAF-MAPK and PI3K-AKT signaling pathways.

Jeffrey Loeb (University of Illinois at Chicago) discussed mapping seizures in SWS and integrating clinical, neuroimaging, and histological data.

Barbara Handelin (Rare Disease Alliances) introduced the Rare Disease Alliance and its goals.

Kellie Sadens (PEN) led a joint PEN / SWFIRN discussion on issues relevant to both groups. For many of us involved in basic research this provided a welcome focus on research issues relevant to patients and their families. See the conclusions below.

Sergiusz Jozwiak (Children's Memorial Health Institute, Warsaw) discussed preventative epilepsy treatment in SWS and tuberous sclerosis.

Thuy Phang (Baylor College of Medicine) discussed the establishment of an endothelial cell line from a SWS brain sample and characterized its proliferative and other properties.

Csaba Juhasz (Wayne State University School of Medicine) described multimodal imaging to understand brain abnormalities in children with SWS.

Kiersten Ricci (Cincinnati Children's Hospital Medical Center) reported that sirolimus appears efficacious in reducing symptoms and improving quality of life in patients with Klippel-Trenaunay Syndrome (KTS).

J. Michael Taylor (Cincinnati Children's Hospital Medical Center) discussed MRI-based arterial spin labeling in SWS as a way to differentiate SWS from isolated facial capillary malformation in early infancy.

Leslie Garson (UC Irvine) discussed anesthesia for laser procedures for port wine birthmarks and other vascular anomalies.

Anna Pinto (Boston Children's Hospital) presented on brain imaging in patients with Sturge-Weber syndrome and presenting with acute neurological symptoms.

Adrienne Hammill (Cincinnati Children's Hospital Medical Center) discussed a classification of vascular anomalies, including vascular malformations (present at birth) and vascular tumors, highlighting a series of syndromes seen at the Hemangioma and Vascular Malformation Center at CCHMC.

Anne Comi (Kennedy Krieger Institute) discussed presymptomatic treatment of SWS using aspirin and anticonvulsants.

We welcomed many other participants such as **Jack Arbiser** (Emory University School of Medicine) and **Scott Mellis** and **Giusy della Gatta** (Regeneron).

Doug Marchuk (Duke University School of Medicine) and

Jonathan Pevsner (CSO, SWF) summarized the meeting by discussing priorities for the SWS research community.

Conclusions (part 1): patient-oriented issues. Future

directions for SWS (and for SWF support of SWS research) include the following:

- We need to understand better the natural history of SWS
- We need to complement pediatric studies with adult issues (e.g. why adults have seizures; changes in vascular abnormalities over time; migraines; mental health issues; social issues relevant to adults)
- Focus more on quality of life issues for individuals with SWS
- Continue to focus on optimizing existing treatments, and introducing new treatments (from laser therapy to gene therapy)

Conclusions (part 2): research directions (areas the SWF can offer direction and support):

- Understand disease pathogenesis
- Introduce standard nomenclature
- Introduce a biobank
- Have the SWF support researchers through seed grants.
- Have the SWF support researchers through grants for young investigators
- Support NIH funding for SWS research
- Develop an Institutional Review Board (IRB) protocol and consent form to facilitate sample acquisition (e.g. from biopsies) and further studies such as establishing cell lines and genomics
- Support creation of relevant databases
- Support genomics efforts
- Support development of mouse models and other animal models
- Support the patient registry

These conclusions about research needs represent the perspectives of patient and family needs; basic research needs; and applied clinical research needs. The Sturge-Weber Foundation looks forward to actively pursuing these ideas and supporting forward progress in a collaborative manner. The SWF is prioritizing these suggestions based on available resources. Please direct any comments, questions, or requests to Jonathan at cso@sturge-weber.org or phone (443)923-2686 (office).

Sincerely,

Doug Marchuk, Ph.D.
James B. Duke Professor and Vice-Chair
Department of Molecular Genetics and Microbiology
Duke University Medical Center

Jonathan Pevsner, Ph.D.
CSO, The Sturge-Weber Foundation
Professor, Dept. of Neurology
Kennedy Krieger Institute and Johns Hopkins Medicine

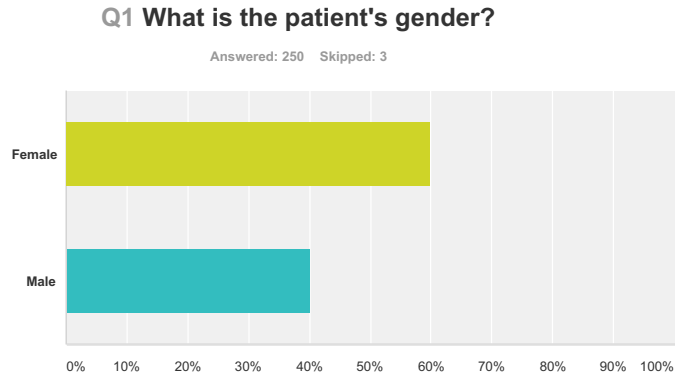
THE STURGE-WEBER FOUNDATION

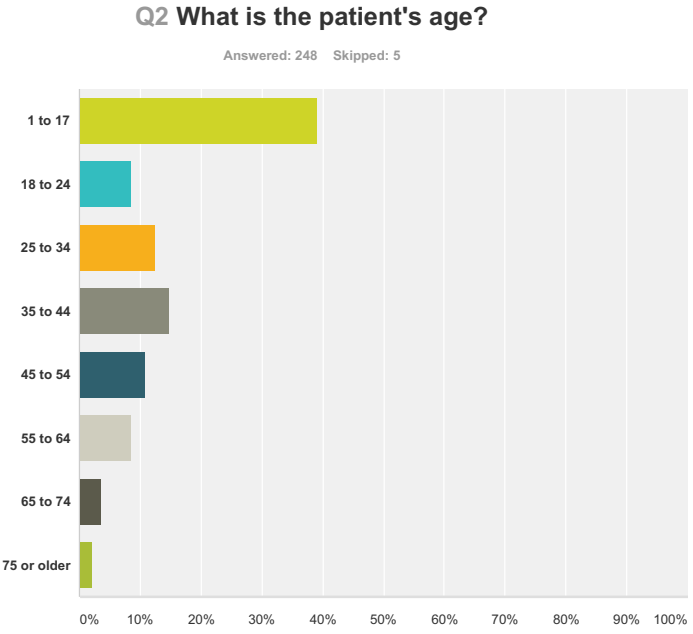
PATIENT ENGAGEMENT NETWORK

PATIENT ENGAGEMENT NETWORK (PEN)

The Patient Engagement Network (PEN) brought patient advocates from the USA, Canada and Puerto Rico to develop an engagement plan and survey to distribute to patients. The following summarizes the results of 253 respondents.

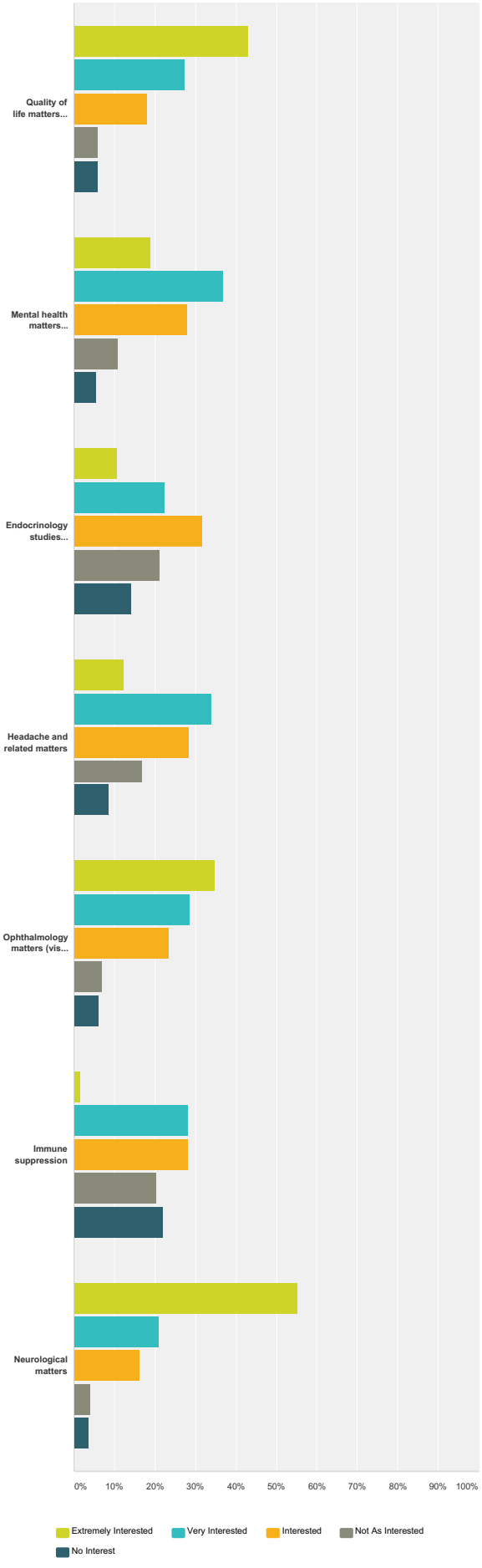
We appreciate and value your input via the website, social media, and email.





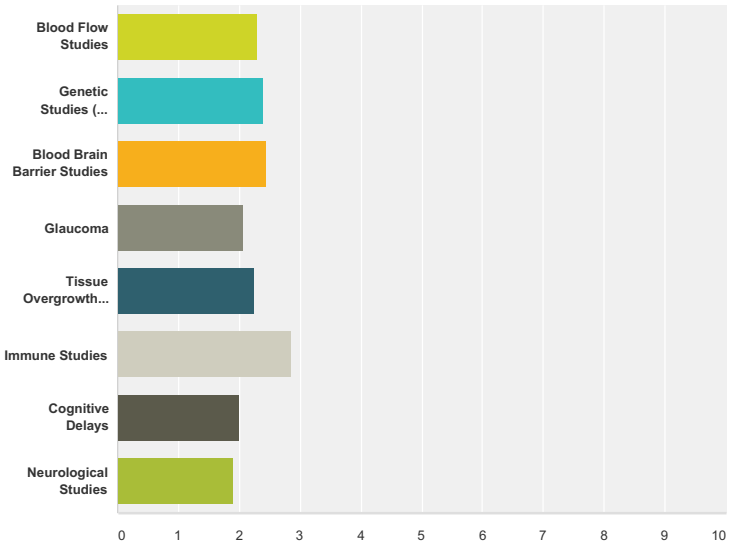
Q6 Please rank the top three areas of interest for clinical research (1-6):

Answered: 247 Skipped: 6



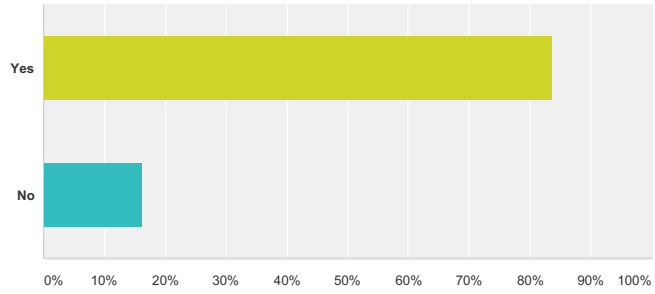
Q7 Please rank your top three areas of interest in basic research:

Answered: 243 Skipped: 10



Q18 Would you be willing to sign a consent form to participate in clinical research (blood draw, saliva sample, biopsy)?

Answered: 220 Skipped: 33



LISA'S RESEARCH FELLOWSHIP

With the support of Lisa's Sturge-Weber foundation fellowship, Dr. Huang published a paper demonstrating that *GNAQ* mutation is primarily carried by the endothelial cells in brain capillary malformations in Sturge-Weber syndrome (SWS). This study, combined with her previous publication on skin capillary malformation, points SWS researchers to *GNAQ* mutation-bearing endothelial cells in capillary malformations and provides a critical starting point for cell-based studies.

Brain tissues from 2 individuals with SWS were obtained from neurosurgical procedures. Dr. Huang isolated *GNAQ* mutation-carrying endothelial cells from these specimens in the laboratory. She found the *GNAQ* mutant endothelial cells behaved abnormally compared to normal brain endothelial cells when the cells were stimulated with vascular endothelial growth factor (VEGF). VEGF is an essential growth factor that regulates endothelial behavior and function, and it stimulates capillary blood vessel growth. The altered behaviors induced by VEGF may help to explain the abnormal capillary blood vessels in SWS brain.

One way cells communicate with neighboring cells is through secreted proteins. Therefore, Dr. Huang analyzed the proteins secreted by *GNAQ* mutant brain endothelial cells versus normal brain endothelial cells. She found 20 different proteins that were secreted at either increased or decreased levels when compared to normal brain endothelial cells. She is now testing whether some of these proteins may be involved in miscommunication between blood vessels and neurons, which could be a contributing factor to SWS. This project is being conducted in collaboration with Dr. Mustafa Sahin's laboratory at Boston Children's Hospital.

GIVE THANKS
AND A WONDERFUL PRESENT BY UPDATING OR STARTING YOUR REGISTRY PARTICIPATION TODAY!

STURGE-WEBER INTERNATIONAL REGISTRY

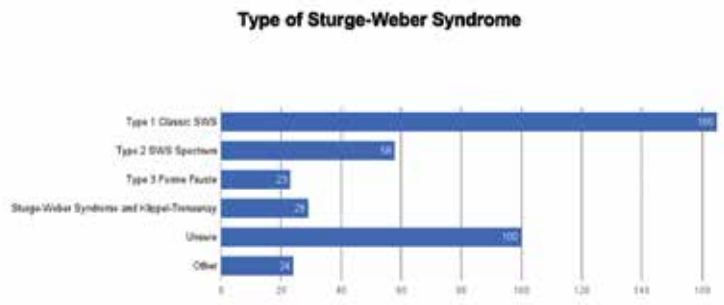
www.sturgeweberregistry.patientcrossroads

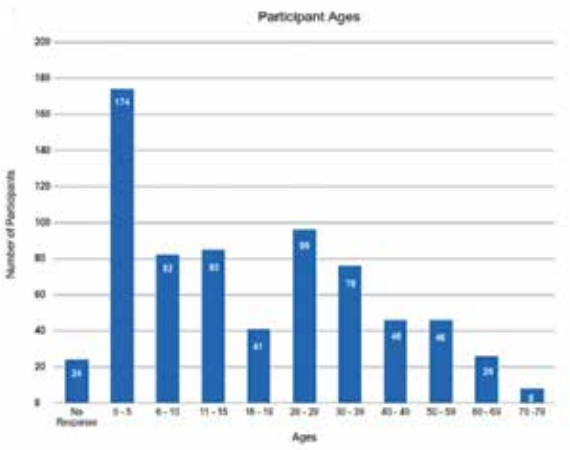
The SWS community has greatly benefited when you reach out and share your stories and medical concerns. Not only does it add to the collective knowledge base of the natural history of the progression but this collective data has enabled our scientists to investigate and identify emerging aspects in need of further study...as well as finding the *GNAQ* gene mutation!

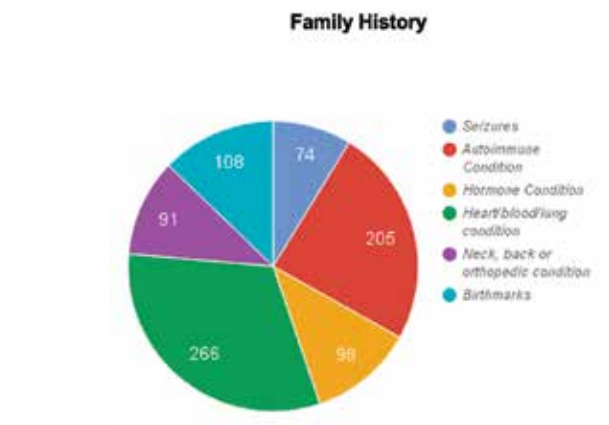
RONNIE AND JAKE

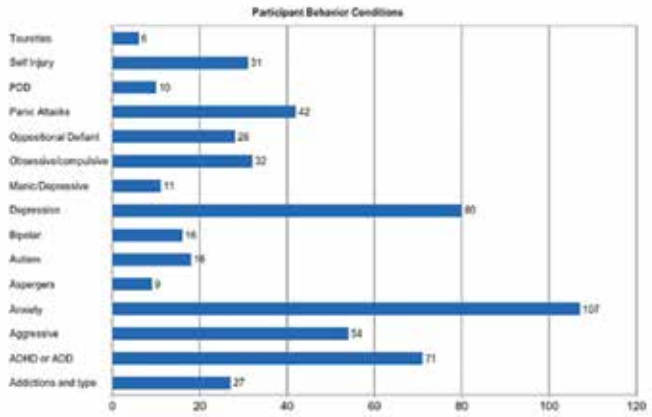
Why Join?

- YOU can help researchers better understand the impact of living with SWS - in a way that ONLY patients can.
- YOU can join a program that puts your privacy & security first.
- YOU can see how your responses compare with others.
- YOU can help to identify gaps where treatment is most needed.
- You can help researchers understand differences in disease severity.
- YOU can accelerate development of new treatments.









BRAIN VASCULAR MALFORMATION CONSORTIUM (BVMC)

Brain Vascular Malformation Consortium (BVMC) received a second round of funding from the NIH. The BVMC is a collection of academic medical centers, patient support groups, and clinical research resources dedicated to conducting clinical research on different forms of brain vascular formations, including Sturge-Weber syndrome (SWS). The Sturge-Weber Syndrome Project of this grant is led by Dr. Anne Comi, MD at the Kennedy Krieger Institute, Doug Marchuk Ph.D. at Duke University, and Karen Ball at the Sturge-Weber Foundation. Its goal is to better understand the clinical issues in Sturge-Weber syndrome and develop new tools for monitoring and treating the brain, skin, and eye conditions associated with the syndrome.

The study has three aims: 1) develop a national database for SWS and determine how changes of the port-wine birthmark and eye are related to brain and nerve function and levels of certain vascular factors found in the urine, **2)** assess changes in deep draining blood vessels in SWS as it relates to stabilization of brain and neurological functioning using brain neuroimaging scans, and **3)** explore changes in protein pathways as a result of the SWS-causing *GNAQ* mutation using skin and brain tissue samples.

National De-Identified Database: We have reached almost 300 participants and began enrollment with the help of the SWF and the other six recruiting sites.
Other projects underway at select sites include:

- **Urine Biomarker Development:** We are recruiting at several sites..
- **Neuroimaging:** Neuroimaging scans provide a detailed view into the inner workings of Sturge-Weber syndrome brain involvement. We continue to actively recruit for the prospective neuroimaging aspect.**Tissue Sampling:** In skin and brain tissue of those with Sturge-Weber syndrome, there is a mix of mutant and non-mutant cells. We begann enrollment of those with port wine stains (PWS), collecting both PWS tissue and non-PWS tissue.
- **Pilot Project:** The SWF's Center of Excellence at Cincinnati Children's and the Kennedy Krieger Institute are collaborating with the BVMC DMCC to study sirolimus for cognitive impairment in SWS.

A LASTING LEGACY AND GIFT: BRAIN AND TISSUE DONATION

Maryland Brain and Tissue Bank/NIH NeuroBioBank As of the fall of 2014, over 900 researchers in 26 countries had received over 35,000 tissue samples. These researchers have published about 700 scientific papers based on research with tissue donated to the Bank. Approximately 50 disorders have been studied.

In 2013, three NIH Institutes (NIMH, NINDS and NICHD) decided to enhance the value of brain and tissue banks for both the tissue donors and the researchers. To this end they funded the **NIH NeuroBioBank** (neurobiobank.nih.gov) which is a network of 6 brain and tissue banks with the directive to collect tissue from brain and tissue donors of research interest to all three institutes.

Tissue will be housed at the bank that collects the tissue. All tissue requests will be made on the NIH NeuroBioBank website. A central committee will review tissue requests. An individual or several banks will indicate if they can fill the request. One common Material Transfer Agreement will be used by all 6 banks. Donor information will be standardized. Standard quality standards will be instituted. In 2014 the contract supporting the **University of Maryland Brain and Tissue Bank** was renewed under the auspices of the NIH NeuroBioBank. This bank will adhere to the guidelines of the NIH NeuroBioBank by collecting a broader range of disorders, however, special emphasis will be placed on developmental disorders, especially Autism Spectrum Disorders.

The emotional and physical well-being of the families and individuals we interact with is our greatest priority. Time is taken to inform everyone involved of the purpose and process of tissue donation. All questions are answered to

the best of our ability. Written information is provided to potential donors for future reference. All donor information is kept in strictest confidence. We attempt to instill in all professional staff the importance of the great contribution which donors are making to society.

Since every effort is made to encourage professional staff to volunteer their time and effort on behalf of medical research, we acknowledge sincere gratitude to all individuals participating in tissue retrieval.

Representatives of the Bank will make every effort to inform qualified researchers of the availability of the tissue. In doing so, the goal and purpose of the precious gift of tissue can be fully realized.

Every effort will be made to assure the equitable distribution of tissue, with special emphasis on furthering research directed toward improved treatment or a cure for the disorder of the donor.

Tissue recovery occurs at the most stressful time for the family. As the recovery teams are under a time constraint to obtain the tissue in optimal condition for research, full appreciation has to be given to the fact that misunderstandings may arise. A great deal of forbearance has to be shown by all individuals involved.

We are dedicated to realizing fully the thoughts and hopes of the individuals and families who contribute these precious gifts for the benefit of all.

TO DONATE SKIN, BRAIN OR EYE TISSUE, PLEASE CONTACT:

UNIVERSITY OF MARYLAND BRAIN & TISSUE BANK
A Brain and Tissue Repository of the NIH NeuroBioBank

Dr. Ron Zielke
NICHD Brain and Tissue Bank
University of Maryland
655 W. Baltimore St
Baltimore, MD 21201
800-847-1539, 419 706-1733
Btbankfamily.org

DR. SAHIN AND DR. PINTO AT THE BOSTON CENTER OF EXCELLENCE

SWF CENTERS OF EXCELLENCE (COE)

SWS CENTER OF EXCELLENCE
THE STURGE-WEBER FOUNDATION

You deserve to have experts in SWS, KT and birthmarks healthcare and research as close to home as possible. Gone are the days where you need to have out of pocket additional expenses to fly across country to receive topnotch care or to participate in research. To that end, the SWF has been visiting with experts in diverse specialties to engage in the exciting research you are reading about in this issue and more in the planning stages. We are excited to share with you the latest COE's and contacts. Anne Howard can still assist you with reaching out or answering your personal questions but we encourage you to check in frequently to keep up to date on all the COE news.

CA: **UC Irvine Beckman Laser Institute – Irvine**
CA: **UCSF Medical Center – San Francisco**
CA: **Rady Children's Hospital – San Diego**
DE: **Nemours/DuPont Hospital for Children – Wilmington**
IL: **Un of Illinois at Chicago Medical Center – Chicago**
IL: **Lurie Children's Hospital – Chicago**
MA: **Boston Children's Hospital – Boston**
MI: **Children's Hospital of Michigan – Detroit**
MN: **Mayo Clinic – Rochester**
Satellite Clinic in Phoenix, AZ
Satellite Clinic in Jacksonville, FL
NC: **UNC Children's Hospital – Chapel Hill**
NJ: **Northeast Regional Epilepsy Group – Hackensack**
NY: **NYU Medical Center – NYC**
OH: **Cincinnati Children's Hospital – Cincinnati**
OH: **Nationwide Children's Hospital – Columbus**
PA: **Thomas Jefferson Univ. Medical Center – Philadelphia**
PR: **Centro Medico de Puerto Rico – San Juan**
TX: **Dell Children's Medical Center – Austin**
TX: **Texas Children's Hospital – Houston**

OVERVIEW OF FEDERAL ACTION FOR THE STURGE-WEBER FOUNDATION

Lobbyit was engaged by the Sturge-Weber Foundation to be their representative in Washington. Lobbyit is working on three main areas of advocacy:

1. Advocating to increase research funding
2. Raising awareness of Sturge-Weber
3. Organizing a "Hill Day" (May 2017) and briefings on the Hill

Washington at a Glance:
On November 8th, Donald J. Trump was elected the 45th President of the United States, and has a Republican majority in both the Senate and House with which to govern. Having a single party in charge of the legislative and executive branches of the government often reduces gridlock and can expedite legislative priorities. One caveat is that there is a "magic number" of 60 votes needed in the Senate to get anything done. While Republicans maintained their majority in the Senate, they lack the 60 votes needed to push through legislation and will have to rely on at least 9 Democrats to move legislation through the Senate. The House is still solidly Republican and does not have the same issue as the Senate. This forces the legislative agenda to be more moderate than it otherwise would be when one party is in charge of the executive and legislative branches.

In the health care and medical research realm, President Trump has promised to overhaul/ ditch the Affordable Care Act, what the replacement looks like is uncertain at this point. Medical research at the Department of Defense, which allows researchers to have higher risk factors in their project than the NIH prefers will continue unaffected. With a new President and Congress, this affords the Sturge-Weber Foundation a perfect opportunity to weigh in, make their voices heard and advocate for research dollars within the federal budget.

Legislation of interest:
There are several bills that are of great importance to rare diseases in general and Sturge-Weber in particular. Everything that has not gone to the President's desk for signature at the end of the calendar year expires and needs to be re-introduced during the next Congress, which starts January 2017.

First is the 21st Century Cures legislation. The Cures legislation:

- Requires NIH to establish a national pediatric research network and pool resources and coordinate activities related to pediatric rare diseases
- Establish public-private partnership to establish and maintain a database on history of diseases with a focus on rare diseases
- Incentivize repurposing of major market drugs for rare diseases
- Reauthorize the rare pediatric disease priority review voucher (PRV) program and broaden the definition

This legislation passed through the House and is awaiting Senate action. It has been identified as a top priority for the lame duck (post-election) session, and President Obama has expressed a willingness to sign the legislation into law if it passes both the House and the Senate. Lobbyit is advocating for passage of 21st Century Cures on behalf of the Sturge-Weber Foundation during the fall. This bill is a top priority and one of the most important pieces of legislation for rare diseases this year.

Next up is the National Defense Authorization Act (NDAA) and defense appropriations. These two are required every year to fund our nation's armed services. In addition to tradition vehicles, weapons, personnel there is a great deal of funds earmarked for medical research. The research must be tied to the welfare of service members and their families, but offers additional opportunities for researchers that may not have advanced their research far enough along for NIH grants. The NDAA creates the programs, such as the Congressionally Directed Medical Research Program (CDMRP) and the defense appropriations bill funds those

programs. Neither the NDAA nor the defense appropriations bill has been sent to the President's desk for signature, however both must be completed by the end of the year to ensure our nation's military is adequately funded. Lobbyit is advocating on behalf of the Sturge-Weber Foundation for passage of the defense appropriations bill with funding for rare disease research.

Another important piece of legislation is S. 2188, the Rare Diesease Innovation Act. This bill was introduced by Senator Cory Gardner (R-CO) and it:

- Amends Federal Food, Drug and Cosmetic Act to expand humanitarian device exemption to authorize the FDA to exempt from effectiveness requirements medical devices intended to benefit fewer than 8,000 individuals (currently the number is 4,000).

This can expedite the process of approval for medical devices required to treat rare diseases. Lobbyit is currently following this legislation, as action does not appear imminent.

Caucuses and Related Organizations:

Lobbyit began work with the National Organization of Rare Diseases (NORD) on a collaboration to advocate for passage of 21st Century Cures. NORD's legislative priorities for the next year are largely at the state level, but at the federal level, their top priorities are 21st Century Cures and the reauthorization of the Prescription Drug User Fee Act (PDUFA).

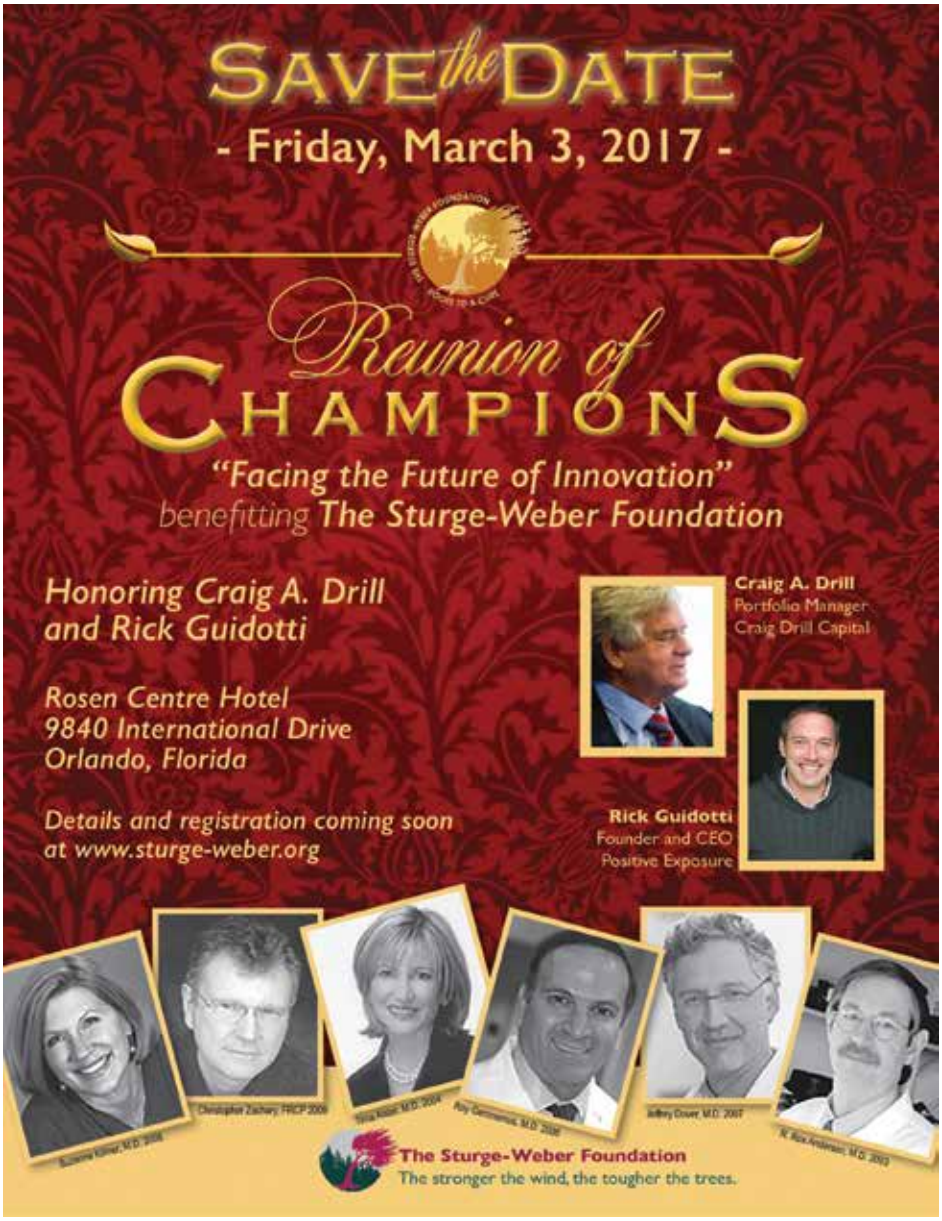
Lobbyit has begun working with the Rare Disease Congressional Caucus co-chaired by **Sen. Klobuchar** (D-MN), **Sen. Hatch** (R-UT), **Rep Crowley** (D-NY), and **Rep Lance** (R-NJ). The Caucus often takes the legislative lead on rare disease issues and is vital to new rare disease legislation. Lobbyit has introduced the caucus to the Sturge-Weber Foundation and formed a working relationship that will benefit the foundation and efforts to secure additional research dollars.

Furthermore, the Foundation's Congressional delegation of **Rep Frelinghuysen** (R-NJ) and **Rep McCaul** (R-TX) are vital to our efforts.

Call to Arms:

None of these items will achieve their maximum effectiveness without the efforts of patient advocates. What matters most to Members of Congress and their staff are their constituents' stories.

Lobbyit encourages all patient advocates to reach out to **Brandon Farris** (bfarris@lobbyit.com) to discuss their Congressional delegation and how they can advocate for additional research funding. We need your help to make these efforts a success!



OFFICE SPACE

Actually, that is a misnomer. The SWF no longer has working space at 1240 Sussex Turnpike in Mt. Freedom NJ. I guess you could say we are in "Outer Space" since we are now a virtual office. The transition was challenging but we are almost settled.

Karen Ball, our CEO is in Aurora, CO; Brian Fisher our VP of Operations and Corporate Relations, is in Houston, ably assisted by Susan Finnell, our Director of Marketing and Programs. Julia Terrell handles our Social Media (and the Cincinnati Conference) from her home office in Sicklervills, NJ and Anne Howard, Patient and Family Services, has a home office in Mendham, NJ where she has access to the PO and bank. We all have phones, computers and printers.

The phone number is the same **973-895-4445** and when you call you will be given a choice of whom you want to speak with. The mailing address is the same – **PO Box 418, MT Freedom, NY 07970.**

MEET SUSAN FINNELL

Susan Finnell, the new Director of Marketing and Programs has a background that fits well with the SWF.

In her own words: I came from Houston Christian High School and was the Marketing and Communications Associate. Houston Christian is a private college preparatory school. I handled advertising, webmaster, graphic design, and development campaigns for the school along with the Marketing Director. Prior to that I worked for West University United Methodist Church and Westbury Baptist Church for 12 years combined as marketing and communications director and printshop manager. Degree in Visual Communications. I am married to Andrew, I have 1 daughter, Katrina, 27 and a step-daughter, Ashley, 30 and step-son, Holland, 36, and 4 grandchildren. Oh, and I am a native Houstonian.

I have worked in the non-profit sector for 17 years, primarily within religious and academic areas. This is my first time to work in the medical/science area, but I find it extremely interesting and love meeting the families, patients, doctors, etc. who support SWF. I have always wanted whatever I did as a profession to directly affect the lives of others in a positive way and I believe I have found that here as SWF.

SAYING 'GOODBYE'

Bonnie Ayers, who first joined the SWF in 2007, leaves behind a vast organized network of fundraising and administrative programs. We will miss her energy. She now has the opportunity to focus on family and projects that will no doubt find their way to her.

The SWF owes Bonnie a big Thank You for building up the fundraising and technical data programs we have now.

ON THE ROAD…

The SWF staff and volunteers give presentations around the country and man exhibit booths to educate and engage attendees. These presentations and interactions create opportunities to identify emerging areas of concern, develop new studies and spread awareness of the SWF and conditions we serve each day.

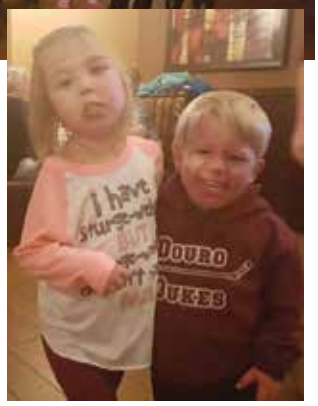
- **Indication Expansion and Drug Repurposing Summit**
  Brian J Fisher speaker
  Washington, DC May 2016
- **Industry & Research targets and seed planting to put Patients in the forefront in research development– Partnering with IDNs BioPharma Strategy Summit**
  Brian J Fisher Moderator
  Washington, DC August 2016
- **Industry and commercial seed planting of Patient Advocacy as a Partner – Strategic Partnerships for Repurposing Forum**
  Brian J Fisher Moderator
  Boston, MA
  October 2016
- **Industry & Research targets and seed planting to put Patients in the forefront in research development–Child Neurology Society**
  Karen L Ball Exhibitor
  Vancouver, Canada October 2016
- **PCORI Annual Meeting**
  SWF Represented as participants
  Washington, DC November 2016
- **Cannabis-Based Therapies Conference**
  Brian J Fisher Speaker
  San Francisco, CA November 2016

ROCKIN' TOWARDS A CURE

Kris and Kellie Sadens hosted a family Karaoke night in November at the Rock House in Glenview, IL for about 20 families. Including a match from Hulu, the donations totaled over $2700. *Rock On!*

Kellie says, "A doctor told us early on that Silas might not develop an appreciation for music because of the area of his brain that was affected by SWS. However, in true rock fashion, Silas ignored the doctor and has grown up to be a passionate music fan of his dad's favorite classic rock and his mom and sister's penchant for pop. Our whole family loves Friday Karaoke night at the Rock House and we invited our friends and family, along with SWF's Emily Argersinger and Tynan Benson and families, for an evening to celebrate defying the odds and embracing your inner rock star".

FRIENDRAISER IN CANADA



Candice Roberts and her family hosted a Spaghetti Dinner Friendraiser in their small but mighty town of Napanee, Ontario. Candice reports that there was a huge storm just before and she was worried they might lose power but that did not deter Harley's friends and family. As usual, "Harley's Army" showed up in force. The dinner was held at Crabby Joe's and they served over 90 plates of spaghetti at the friendraiser in addition their regular business that night.

Nicole Deutekom and little Max and the rest of their family came all the way down from Peterborough in the storm. It was a busy, exciting and heart -warming night.

"We were able to raise over $1700 for the SWF. We are planning a baseball tournament this summer as our second Friendraiser."

RHODE ISLAND DAY

Annette Coutu, grandmom of Brielle, planned, invited and hosted a get together at Fort Wetherill in Jamestown, RI for a SWS Family Get Together Day. Although rain was in the forecast families still met up to share stories and enjoy a picnic lunch. Emily (age 15) and Brielle (age 21 months) hit it off right from the start. Emily has Type 2 SWS and Brielle has Type 1. Although a small turnout, plans are in the works for another R.I. SWS Family Get Together summer 2017 in hopes to bring more families together.

DUXBURY DOES IT AGAIN

The Duxbury (MA) High School girls volleyball team, the Duxbury Dragons, chose the SWF for their annual benefit game for the second year, raising over $1500.

REPRESENTING US

Cy Simonsgaard and Alex Walsh, members of our Grassroots Advocacy team, attended the CSD (Coalition of Skin Diseases) Development Day in Washington, DC on September 11. Alex and Cy both agree it was extremely informative and wonderful to get to meet other advocates with such a diverse array of rare conditions and

Continued on page 17...

Members Moving Us Along continued...

learning the different techniques in making an impact on legislative visits. Alex shared her handouts and notes with us. Thank you ladies.

GROWING INTERNATIONALLY

We have word from Ana Garcia de Polevieja that there is now an official association in Spain with 20 families and growing. This includes 17 children and 3 adults, with one recent hemispherectomy surgery with great results. They are planning a medical conference in Spain next year at the children's hospital in Madrid. That is just the beginning of their projects and they are eager to spread the word.

Ana Garcia de Polevieja is president of the Asociacion Espanola Sindrome de Sturge-Weber, info@sturge-weber.es.

ASHLEY AND FAMILY

Ashley Sheffer turned 21 in August and graduated from morgan High School in Utah..

BAIILEY AND MOM

Bailey Ellenson in North Dakota turned 18 in April.

Remembering Loved Ones

[WE HONOR NOT JUST THEIR LIVES, BUT THE LEGACY THEY LEFT.]

Vida Garcia-Infante passed away on August 28, 2016, her 2nd birthday. Vida's aunt Karina had contacted us after she was born to provide information, support and help for her sister Rosanna. Throughout Vida's short and precious life, the family was comforted and encouraged by the SWF friends who reached out. Friends and family surrounded them with generosity through a Go Fund Me page that donated more than $7000 to the SWF so we can continue to provide the same kind of caring to other families.

Emily Argersinger's grandfather Brian Earle Fingerle, died in August at the age of 81. He was a veteran of the US Army.

EDUCATION FORUM IN BOSTON

Boston Children's Hospital hosted a SWF Education Forum on November 12 at the Deveber Conference Room in Boston Children's at Waltham.

Families and adults attended to hear developments in the progress towards optimum treatment and care, share their personal journeys and be able to dialog and present their questions and concerns with the speakers.

Brian Fisher, SWF VP greeted the audience and Katherine Keever, BCH Clinical Research Coordinator presented the groundwork "Who We Are and What We Do". Speaking specifically on Research were **Mustafa Sahin, MD,** co-director of the SWS Center of Excellence, **Joyce Bishoff, PHD** of the vascular biology program, **Lan Huang, PhD** of the Bischoff Laboratory.

Masanori Takeoka, MD outlined seizure management in SWS while **Anna Pinto, MD** co-director of the SWS Center of Excellence, spoke on clinical management. **Katrina Boyer, PhD** and **Kitty Perry, M. Ed** covered the topics of neuropsychology assessment and Special Education.

We are grateful to **Pam McIntyre**, SWF Board Chair, for planning this event and to **Katherine Keever** of BCH for getting the details in hand. Boston Children's Hospital has been one of our prime Centers of Excellence and rightly deserves the recent designation of "Best Children's Hospital" by US News and World Report.

BEFORE YOU KNOW IT...

The SWF 2017 Reunion of Champions, which occurs in conjunction with the annual AAD (American Academy of Dermatology) meeting, will be held **Friday, March 3** at the Rosen Centre Hotel in Orlando, FL. This event will honor Craig Drill, a long time benefactor of the SWF and Rick Guidotti, Founder and CEO of Positive Exposure, who is well known to our SWF families. Rick's work as a photographer has graced our publications and enlivened our International Conferences since forever.

Reunion of CHAMPIONS 2017

INTERNATIONAL CONFERENCE IN JULY IN CINCINNATI

All information is on website.

WHEN YOU ARE CHATTING: remember not every Facebook page that has Sturge-Weber in its title comes from us – we have an official general page – **THE STURGE-WEBER FOUNDATION** – and a private (secret) page only for SWF members who ask to be in – **WORLD OF CARE AND SHARE NETWORK.** Spreading the word is great – but only the messages on those two pages are sent by the SWF and come to the SWF.

Take time to remember . . .

We are blessed that you are a SWF family member!

Your contributions of time, talent or treasure have helped the Sturge-Weber Foundation continue to provide assistance to newly diagnosed patients and their families, continue important research, build relationships with Centers of Excellence throughout the United States and build awareness of SWS to the public. We are so grateful to you for helping to create and support SWF.

You are helping SWF touch the lives of thousands seeking to improve their quality of life.
We exist to serve you and your loved ones – to provide hope, healing and a sense of community.
Your gift is extremely important because it offers immediate resources that are directed to current needs and opportunities.

As this year draws to a close we continue to strive towards more medical advances, awareness, partnerships for you, for your loved ones, for every person living with SWS. Please help to make this possible by considering a year-end donation to SWF. Your donation is helping to find roots to a cure, right here, right now.

Happy Holidays from the Sturge-Weber Foundation

The stronger the wind, the tougher the trees

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2017 REUNION OF CHAMPIONS
ORLANDO, FL

JULY 2017

INTERNATIONAL CONFERENCE
CINCINNATI, OH
JULY 26-29, 2017

THIS ISSUE IS BROUGHT TO YOU IN PART THROUGH THE GENEROUS CONTRIBUTION OF THE ALLERGAN FOUNDATION.

"ENJOY THE FRUITS OF YOUR LABORS THIS SEASON."

KONTUR KIDS

IF YOU HAVE OPINIONS, QUESTIONS, OR ARTICLES FOR BRANCHING OUT, WE WOULD LIKE TO HEAR FROM YOU. PLEASE FAX, MAIL, OR E-MAIL YOUR MATERIAL TO THE SWF OFFICE (SWF@STURGE-WEBER.ORG). WE RESERVE THE RIGHT TO EDIT ALL MATERIALS.