



The Sturge-Weber Foundation
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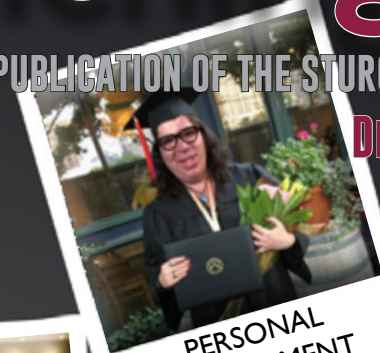
**A LIFELINE FOR
A LIFETIME.**



Branching Out



A PUBLICATION OF THE STURGE-WEBER FOUNDATION
DECEMBER | JANUARY



**PERSONAL
ACHIEVEMENT**



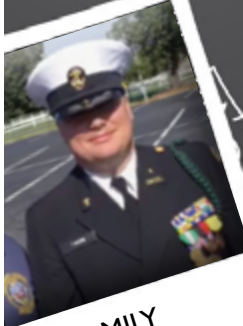
**CELEBRATING
CAREGIVERS**



**EVENTS &
CONFERENCES**



**MONTH OF
AWARENESS**



FAMILY



**FALMOUTH
ROAD RACE**



**LIFELINE FOR
A LIFETIME**

**ROOTS TO
RESEARCH**

**2019 INTERNATIONAL
FAMILY CONFERENCE**

**SPOTLIGHT
ON RESEARCH**

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The SWF is a member of the Brain Vascular Malformation Consortium (BVMC), American Brain Coalition (ABC), The Coalition of Skin Diseases (CSD), and the Association for Research in Vision and Ophthalmology (ARVO).

Branching Out

December/January
2018-2019

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Thank you for your
continued support through
Best Local Charities of
America (SWF #11857).

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).

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 a clearinghouse of information, provide emotional support and facilitate research on PWB and SWS.

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 STURGE-WEBER FOUNDATION
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NETWORK

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- **UC-Irvine**
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- **UCSP Medical Center-San Francisco**
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Delaware:

- **Nemours duPont Hospital for Children-Wilmington**
Primary Contact: Carol Roethke, CRNP-APRN

Florida:

- **Bascom Palmer Eye Institute-Miami**
Primary Contact: Kara Cavuto
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- **Ann and Robert H. Lurie Children's Hospital**
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- Akira Yoshi, MD | Neurology

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- **Boston Children's Hospital**
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- **Children's Hospital of Michigan-Detroit**
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- **UNC Children's Hospital-Chapel Hill**
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- **Northeast Regional Epilepsy Group-Hackensack**
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- **NYU Medical Center-NYC**
Primary Contact: Daniel Miles, MD | Neurology

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- **Cincinnati Children's Hospital**
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- **Nationwide Children's Hospital-Columbus**
Primary Contact: Warren Lo, MD | Neurology

Puerto Rico:

- **Centro Medico de Puerto Rico-San Juan**
Primary Contact: Rafael Rodriguez Mercado, MD | Endovascular

Texas:

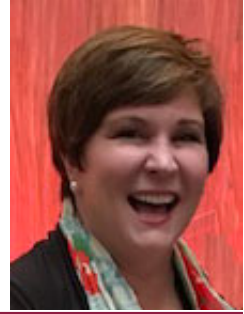
- **Cook Children's Medical Center-Fort Worth**
Primary Contact: Amy Davis, RN
- **Dell Children's Medical Center-Austin**
Primary Contact: Moise Levy, MD | Dermatology
- **Texas Children's Hospital-Houston**
Primary Contact: Denise Metry, MD

Washington:

- **Seattle Children's Hospital**
Primary Contact: Jonathan Perkins, DO | Vascular Clinic Director



Connecting with Karen



For a Reason, a Season or a Lifetime

I was just updating my insurance policies and I had a lightbulb moment! They happen on occasion whenever I get nostalgic and think of the future.

Do you have a life insurance policy? A Will? A Medical Power of Attorney? Special Needs Trust? Most of us, I sincerely hope, have these vital documents safely tucked away in our fire boxes for a rainy and hopefully long into the future day of need. Life with a loved one diagnosed with Sturge-Weber syndrome can be tumultuous with brilliant moments of cherished celebrations for milestones reached.

I've answered thousands of emails and telephone calls from many of you over the years. It's been great to not only catch up on your lives but learn of a new manifestation or some symptom that has cropped up that you need some advice to handle it. The sharing is INVALUABLE to our understanding of the progressive nature of birthmarks, glaucoma, seizures, headaches and more!

I am so happy that you have not felt the need for the SWF until something crops up if it does and that you are leading "normal" lives!! The need for the SWF lifeline is greater now than ever before...we have over 2000 adults living with SWS. There are emerging medical, psychological and social concerns that only those of us living with them on a daily basis can provide insight and hope.

So, here's my request...I am asking you to take out a life insurance policy...not the typical kind you're thinking of, but a small investment that will become an endowment to enable the SWF to be your lifeline when you may need it or to honor those who are on the frontlines still battling seizures, glaucoma and isolation everyday!

We have over 6,000 cases of individuals with SWS, KT and Birthmarks. I'm asking you to be one of the 3,500 people who we no longer hear from until a crisis occurs to step up and give just \$10 as your personal SWF life insurance policy. Just imagine what answers through research and one to one support we could muster up with each one of you giving just \$10. A rainy day policy to ensure we remain on the frontlines fighting for you and yours!

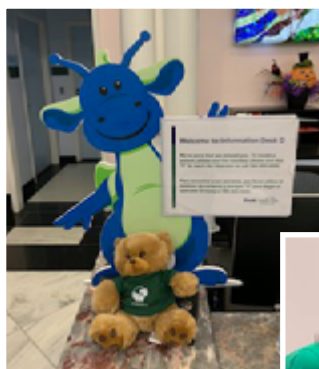
There WILL be a day when my loved ones will have to dust off my life insurance policies. But just for today, you and I can ensure the SWF and our expanding network of care providers will be there to pick up your call to catch up and give you a hand up!

I am thankful to have been a part of your lives these past 32 years and wish you all a very Happy Holiday season! Thank you for considering my request...you ARE cherished friends and we miss you!

Faith, Hope and Love,



RECAP: CCN EDUCATION CONFERENCES



October 20, 2018

Cook Children's Hospital | Ft. Worth, Texas

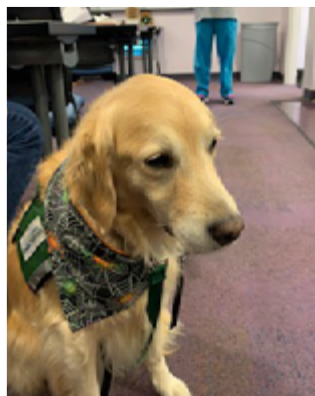
Hosts: Amy Davis, Dr. Shanani, Kia Carter,
Dr. Volkman, Dr. Prashanthi Giridhar

October 27, 2018

**UC-Irvine,
Beckman Institute,
Irvine, CA**

Hosts: Dr. Kristen
Kelly, Dr. Sameh
Mosaed, Dr. Mary

Zupanc, Dr. Nicole Hadley, Michaela Hatch, Dr. Chris Hughes, Andrea Giancarli





Who is ready to getaway for a weekend? We know we are here at the Foundation!

The 16th Annual International Family Conference is coming soon! As one family said about their experience at their first International Family Conference - it was like coming home. It's true, you will meet people just like yourself whether you are a parent, caregiver, family or the patient. You will meet people that are all ages and varying degrees of Sturge Weber Syndrome. The best part though is for 3 days you will be around people that understand what is like...to have Sturge Weber Syndrome.

Some say they are scared to come, some say they don't know what to expect, but everyone says when Saturday night comes and the party is about to end it is bittersweet because you never dreamed you could create a bond so strong. For the first time you know you are no longer alone in your journey!!

So, now that we have convinced you to go, let us tell you about the upcoming 2019 Conference (#2019SWFIFC).

Thursday, July 18 there will be clinics you can meet doctors from many areas of Sturge Weber Syndrome and talk to them one on one about your specific case of Sturge Weber. Our plan is to have doctors from Neurology, Dermatology, Ophthalmology, Behavior Specialist and more. You will be on your own to explore Wilmington, DE and the surrounding area. We are also looking to get together about 7 PM to do a meet and greet so you get the opportunity to meet other families before the conference.

(continued on page 6)



CONFERENCE (continued from page 5)

Friday, July 19, we will begin the day early and head over to Nemours Alfred I. duPont Hospital for Children. This will be a day of learning from all different specialties. We are planning to have two different parts to the conference, one track specializing with newly diagnosed up to teens years, and the second track specializing in adulthood.

Please bring the kids! There will be awesome fun days planned for them also! Once the conference session is over, we will take a walk over to the beautiful Gardens of the DuPont Estate and have the Route to a Cure Walk. The evening will end with a relaxing time back at the hotel.

Saturday, July 20, the conference continues and the bonds will become stronger. At the end of this session, there will be a special event planned for everyone to attend - you won't want to miss it!

So, are you ready to go yet?

If you have never been to one of our conferences you must come and

experience it first hand! If you have come before please come "home" again!!

This area of the country has so much to offer being so close to the museums of Philadelphia, tax-free shopping in Delaware, the beaches and casinos of Atlantic City, NJ and of course not too far from the famous New York City!

If you have suggestions or want to help with the planning please reach out to Julia Terrell at jterrell@sturge-weber.org.

We would love to hear from you.

Now we haven't told you everything. Why? Because this is an adventure, and things are always subject to slight changes!

Check SWF social media sites and emails for updates! In December, look for the registration information in your email box. The Sturge Weber Foundation is so excited to be able to partner with our Clinical Care Network, Nemours Alfred I. duPont Hospital for Children. We can't wait to see YOU this summer!!!

We Love

We Share

We Play

We Laugh

Family

We Fight

We Live



SWF TASK FORCE UPDATE



WHAT IS YOUR PLAN?

Over the last quarter we have seen some great brain storming and ideas come from our newly formed Task Forces. In the new year we will be focusing not only on defining and creating a dictionary of terms you would likely see in each area, but also asking you our members for feedback with surveys and more so that we have a better feel of what is most important to YOU!!! Ultimately creating a place to come when the next phase comes into your world and you will be equipped to create your new Plan. We know that this syndrome is not a straight and easy road most of the time. Instead you have embarked on a journey of many curves, unexpected delays, and well at times just plain chaos leaving us with the feeling of uneasiness. What we learn is it will be ok but wouldn't it be easier with a Plan. As The Task Forces grow we are creating and establishing tools to help that uneasiness turn into a PLAN. We can't control everything but with a PLAN and Tools you will be ready to take on that next challenge with more ease then before. Here is an update on what is happening in each Task Force.

Education Task Force, (Witney Arch, Chairperson) is working on a page on our website to come up with all the ways Education and Transition plays a role in different stages in our lives. Who would be involved and what processes and procedures you need to know about? This year so far we have had 4 very successful Education Conferences where we have learned a tremendous amount of information about Sturge Weber Syndrome and our Clinical Care Networks. Next year we will also be planning even more so look for information in your email and on social media.

Growing Golden Task Force, (Jeffrey Needham, Chairperson) is working on a glossary of terms. What does it mean in this arena to be older and how do you find specialists, employment, and housing? What is the best way to communicate with others?

Growing Up Task Force, (Michelle Daoust, Chairperson) is working on page for our website to include resources and create a portal to have resources available for all areas including community support groups. Information to be included online for all age groups on mental health, services available and where within local communities, suicide helpline, social workers and more. Create chat groups, blogs or podcasts to discuss topics on stages of SWS and what to expect. As we grow we will see this task force and other task forces like Education and Mental Health Task Force will be working together.

(continued on page 8)

SWF TASK FORCE Update



SWF TASK FORCE UPDATE (continued from page 7)

Surgery Task Force, (Candice Roberts, Chairperson) is working on the different kinds of surgeries and creating a glossary of terms. Once completed will look for resources and support groups to help manage as part of your Plan.

Before Their Time Task Force is focused on when we lose the ones we love too soon. We need your help if you are someone that would like to help with this Task Force we would love to hear from you.

Mental Health Programs for Younger Group Task Force, (Donnie Hood, Chairperson) has been focusing on identifying where to find appropriate and accurate resources related to mental health. They are researching websites that are interactive and easy to navigate. They want to include appropriate apps, games and even podcasts while also looking for interventions to improve mental health. They are also working with the Transition Group to develop a possible packet and or forum for both individuals and caregivers. All of this needs to be easy to navigate for the youth population so that we can be sure they have a place to go when the need arises.

Mental Health Programs for Transition Years Task Force, (Kimberly Slater, Chairperson) is finishing gathering the mental health resources and creating a worksheet for reference a cheat sheet so to speak. They are also looking at creating a guide and/or forum for our members to go to when they need help too.

Mental Health Programs for Adults Task Force, (Jillian Barnes, Chairperson) is working on a page for the website to include stories of adults and what they have overcome. Also including resources in both US and Canada (even more as we get them), maybe a discussion board and much more.

Inspire Task Force, (Kellie Sadens, Chairperson) has implemented and successfully ran Ask the Expert. We have planned to do this quarterly so if you have topics or specialties you want to hear more about please let us know. This past quarter we were able to grow with more members joining Inspire to have the ability to have more and more discussions. The wonderful part about Inspire is that these discussions are able to be stored so you can always go back and follow back up on any topic. If you haven't joined please sign up today at swf.inspire.com. In the new year you may even see new Webinars coming our way. Stay tuned.

Nutrition and Complimentary Medicine Task Force, (Stephanie Tikkanen, Chairperson) is an up and coming Task Force. As you all know CBD has been approved by the FDA and we will be looking at this and other alternatives. Stay Tuned.

Grandparents and Care Person Task Force, (Annette Coutu, Chairperson) is looking to create a forum for caregivers and grandparents can go to chat with others like themselves. They have been looking at different possible forums because each group are looking for different things.

Family Day Task Force, (Witney Arch, Chairperson) has been slow in starting only because as the holidays approach it is hard to get together and the hustle and bustle of the season. In the new year we will have a process in place that if you want to have a Family Day we will work with you for invites and the like so we can have Family Days all over the country. These are Fun Days so you get to meet families like yourself. Also being included is events you are planning to support The Sturge Weber Foundation. We will help you come up with a plan to help you promote your special event.

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WHY ENDOCRINOLOGY?

Among the many specialists adults who have SWS see regularly, some are vital and of necessity are seen on a regular basis. Certainly, your neurologist and your dermatologist and often your ophthalmologist. But many adults do not have an endocrinologist on their team, however informal.

Often the role of endocrinology is not apparent in persons who have SWS but there are some physical conditions and body systems that every adult should be aware of.

The most common endocrine condition is diabetes mellitus. Your blood sugar levels should be monitored and treatment instituted if necessary. One of the more prevalent indicators seems to be overweight. Many adults – with or without SWS – can have weight issues, but SWS patients often report overweight issues which is a red flag for diabetes. The opposite is also reported – especially in children. Short

stature and failure to progress along the expected growth curve. This can be treated with human growth hormone and should be monitored if seizure meds are used.

And then any thyroid related concerns, which are often hard to pinpoint because they seem to be so common and “ordinary”, such as fatigue, muscle and joint pain, thinning hair, skin disorders. Thyroid disorders can be hypo- or hyper-. too much or too little. That is why an endocrinology check up is important to determine your status.

Osteoporosis and osteopenia in both men and women should be explored and treated to prevent bone fractures and promote general bone health. An annual DEXA bone scan will keep you alerted to the need for treatment. Vitamin D is essential for the absorption of calcium and should be factored into any treatment plan.

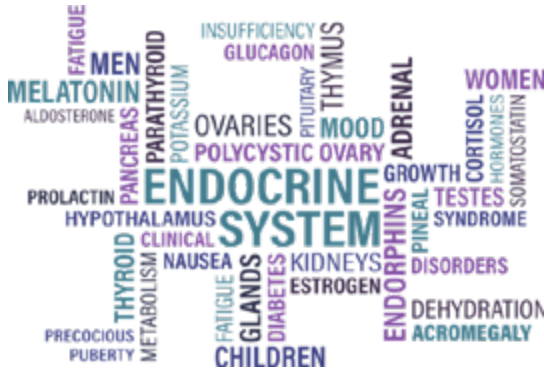
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Some seizure meds are notorious for promoting calcium depletion.

Since many seizure meds have much to do with weight gain, which can lead to diabetes and to cortisol levels, you need a coordinated plan when you are dealing with SWS.



And because puberty (around ages 11-14) is a vulnerable age, boys and girls should be monitored for the many

ways hormones impact development and SWS issues.

SWS patients tend to be very health conscious – and that is a good thing – so these issues are another thing

Menopause in women is related to hormones and should be treated by an endocrinologist as well as a gynecologist. Many SWS patients report that they did not realize why they should be aware of how their endocrine health may be impacted by SWS. Bottom line is that facets of many disorders are sometimes endocrine in nature and should not be neglected.

Not just for adults. Awareness begins early. Many of our pediatric members (kids) have been seen by an endocrinologist because of poor growth or because of overweight.

to put on your check-off list for good health.

But as with all conditions we talk about, ask your primary care physician or your SWS specialist about any health concern and the impact SWS may have on treatment. Do not depend only on Doctor Google for your information. Many internet pages are paid advertisements for medical therapies. The reliable websites are those from medical entities like Mayo Clinic, Massachusetts General Hospital, WEB MD, Cleveland Clinic, Johns Hopkins and other university based medical schools.





YOUR FOUNDATION AT WORK



The SWF Board of Directors and Staff are always at work to bring more awareness, make a bigger impact, seek new opportunities in research and clinical treatments, for the ultimate result - a cure. Here is a brief review of your Foundation's accomplishments during 2018 and a peek at what the future holds.

2018 Accomplishments - Clinical Care

- Begin to finalize big data principles to SWS to identify treatment guidelines
- Continue momentum within CCNs to identify unmet needs and set action points

2018 Accomplishments - Research Roadmap

- Funding for scientific blog to open conversations among professionals
- Expand grants review committee with outside reviewers
- SWF CSO and CCO to participate in major donor visits

2018 Accomplishments - Education and Learning

- Expand online and print support materials each year
- R-13 funding for patient engagement

2018 Accomplishments - Awareness and Advocacy

- Task Forces Implemented

2019 Future Endeavors - Clinical Care

- Develop philanthropic dollars to provide more seed funding for clinical initiatives, new clinical trials and treatments

2019 Future Endeavors - Research Roadmap

- Offer travel grants each year to support travel to post-doctoral fellows, graduate students with specific work in SWS
- Funding for the study of medical data in the SWF Registry and provide analysis
- Coordinate genomics research for the SWS community
- Establish and seek funding for biorepository

2019 Future Endeavors - Education and Learning

- Podcasts with featured experts
- Launch Learning Center with Task Force needs

2019 Future Endeavors - Awareness and Advocacy

- Develop global initiatives for international collaboration

**These accomplishments and future endeavors are possible from the impact SWS patients, families, friends and corporate sponsors have made through faithfully donating to the SWF.
We ARE Better TOGETHER!**

2018



REPORT: ROOTS TO RESEARCH



REPORT: ROOTS TO RESEARCH

The Sturge-Weber Foundation Awards 2018-2019 Lisa's Research Fellowship to Dr. Davide Zecchin University College London

The Sturge-Weber Foundation has named Dr. Davide Zecchin of University College London the recipient of the 2018-2019 Lisa's Sturge-Weber Research Fellowship Award. Research is defined as "patient-oriented research" conducted with human subjects, or "translational research" specifically designed to develop treatments or further various areas of scientific discoveries within Sturge-Weber Syndrome (SWS).

Dr. Zecchin's proposal is "Genetic Therapy for the Sturge-Weber Mutation – Proof of Concept Study." He is an expert in cancer genetics. He received his Ph.D. from the Institute for Cancer Research and Treatment – Candiolo (Turin, Italy) and has performed post-doctoral research in Turin and at the London Research Institute and The Francis Crick Institute. He recently joined the laboratory of Dr. Veronica Kinsler at Great Ormond St. Hospital for Children in London. Dr. Kinsler is expert in mosaic disorders and GNAQ, which when mutated causes Sturge-Weber syndrome. Her lab has shown that GNAQ mutations are responsible for phakomatosis pigmentovascularis (PPV) and Extensive



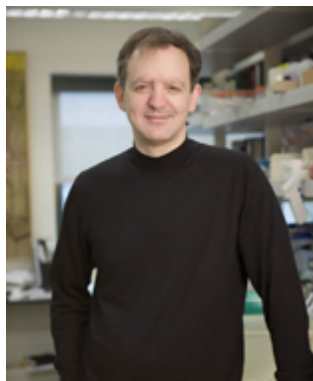
Atypical Dermal Melanocytosis (EDM), and has done pioneering work in characterizing the development of birthmarks.

Dr. Zecchin proposes to demonstrate that Sturge-Weber syndrome (SWS) could potentially be treated by genetic therapy. This genetic therapy would not only be aimed at the skin, but at all GNAQ-mutant cells in the body. The approach is to use gene therapy to correct the GNAQ mutation, working in cells derived from patients. It is thought that gene therapy could be used effectively after birth with the potential to prevent epilepsy, neurodevelopmental delay, and glaucoma.



SPOTLIGHT ON RESEARCH

Dr. Jonathan Pevsner
SWF Chief Scientific Officer



WHAT IS CRISPR?

CRISPR stands for Clustered Regularly Interspaced Short Palindromic Repeats. It's often described along with some associated proteins so it's CRISPR-Cas9. That's a mouthful, so let's just pronounce it "crisper."

Viruses are really interesting things. They tend to be very small—much smaller than a cell in your body. These little viruses can infect cells and cause problems from the common cold to terrible diseases. Viruses aren't exactly living things; they exist on the borderline of the definition of life. They do have DNA (like living things), and they evolve over time, but they can't live on their own.



The tree of life has three great branches (bacteria, other small organisms called archaea, and the eukaryotes that include a range of creatures such as plants, fungi, and animals). It turns out the viruses infect all three branches of life. Bacteria and archaea tend to live as just single cells; but they also have to worry about getting infected by viruses. And they developed a remarkable defense system called CRISPR. CRISPR is basically like an immune system for bacteria (and archaea) to protect them from viruses. Like our immune system there is a "memory" of which viruses attack a bacterium over time.

If you're a bacterium, CRISPR is great. A virus attacks you, and CRISPR fights back: it recognizes the DNA in the virus, attacks it, cuts it, and gets rid of it. And it somehow makes sure NOT to attack the bacterium's own DNA. It's very, very specific.

(continued on page 14)

WHAT IS CRISPR?

(continued from page 13)

Scientists first discovered CRISPR in the 1990s. By about 2013 researchers understood CRISPR well enough to start a revolution in biology. The rate of progress is dizzyingly fast.

What can you do with CRISPR?

Biologists now control CRISPR and use it to edit the genome. That means they can make changes to the DNA. Here is just some of what you can do with CRISPR.

- Take a human cell—say for example a human endothelial cell line growing in a flask in someone's lab. Use the CRISPR machinery to introduce any single change (that's 1 base out of 3,000,000,000). As one example, you can knock out a gene (to study the effects). As another example, you can “turn on a gene.” We can introduce the GNAQ R183Q mutation. Why is that a good thing? We can then test the effects of the mutation on cells, and study whether drugs are helpful.
- You can edit two genes at once. For example, GNAQ and a second gene that we think might interact with it and be important in SWS.
- Instead of cutting DNA to edit a gene, you can “turn on” a gene. (In the language we speak in the lab, we can use CRISPR to recruit transcriptional activators.)
- CRISPR is now used to image DNA in live cells.
- You can use CRISPR “libraries,” as they're called, to search for genes that affect some pathway. This could teach us more about how GNAQ functions with other genes to cause SWS and port-wine birthmarks.

How much can CRISPR help us advance knowledge and treatment of SWS?

CRISPR-Cas9 and related systems are techniques to edit the genome. I see

them as tools that are available today to ask questions about how genes work. I don't see CRISPR tools as leading to an immediate cure for Sturge-Weber syndrome or any other disease. I do see these tools as leading to immediate progress, or at least short-term progress in the upcoming years, to help us better understand GNAQ and how to search for drugs that can be helpful in someday treating SWS.

And the research community is carefully studying the tough question of whether CRISPR actually causes harm. It's possible that it cuts DNA in ways we don't yet understand. For any intervention we balance possible benefit and harm, and with new methods it can take time to figure out possible downsides.

A Goal

CRISPR is amazing. I love reading about tremendous advances being made across a range of areas and especially about diseases. Now let's see some CRISPR advances for Sturge-Weber syndrome.

WHY ANIMAL MODELS?

Animal models have a key role. For genetic disorders such as Sturge-Weber syndrome a change in the DNA (in our case an R183Q mutation in GNAQ) causes the disease in people. If we make the same, corresponding change in GNAQ in an animal then it may get a disease similar to SWS. We can try to figure out how to treat (or cure) the animal, then apply what we've learned to people.

Little Animals

The animals (also called the metazoans) have major branches on the tree of life including worms, insects, and vertebrates. In biology a famous worm is *C. elegans*, and a famous insect is the fruit-fly *Drosophila*. *(continued on page 15)*

WHY ANIMAL MODELS (continued from page 14)

If you visit <http://wormbase.org> and search for GNAQ you'll see that (unfortunately) there's no exact worm version. If you visit flybase.org you can find a fly version of GNAQ (<http://flybase.org/reports/FBgn0004435>).

Sturge-Weber syndrome and capillary malformations are discussed, and there are vast amounts of information about how GNAQ functions in flies (e.g. what else it interacts with and how mutations in GNAQ affect other genes). Flies do have brains and eyes, and as fellow animals they share many features with people. But they're also quite distantly related—we last shared a common ancestor with them about 700 million years ago—so let's take a look for much closer animal models.

Mighty Mouse and Zebrafish

Mice and fish both make great animal models for human diseases. They each have a set of genes that's extremely closely related to human genes. (Who knew!) There are efforts to knock out each of the genes of mouse, and each gene of zebrafish. You can read about this at <http://www.informatics.jax.org/> and at <http://zfinfo.org/>. And all these knockouts and mutants are made available to the research community.

Now, for SWS we want to do something other than knock out a gene: we want to make an activating mutation (it's our familiar R183Q

change in GNAQ) and study its effect. The same mutation in the same gene is a cause of uveal melanoma. Several labs (led by Catherine van Raamsdonk and by Silvio Gutkind) have already made mouse models, published them, and shared their special mice. They focus on other mutations in GNAQ (called Q209L or R183C) and they focus on the effects on other cell types (melanocytes for the melanoma rather than endothelial cells for Sturge-Weber syndrome). But work is well underway to make both fish and mouse models for SWS. We

look forward to several groups reporting their findings very soon.

If you're curious to see information on a GNAQ mouse model go to <http://www.informatics.jax.org/>, enter

GNAQ, and you can find a list of available mouse models [<http://www.informatics.jax.org/searchtool/Search.do?query=gnaq&page=featureList>]. It might look like Greek, but you can see information about a typical mouse model here [<http://www.informatics.jax.org/allele/key/614925>].

So What Do You Do With an Animal Model?

- There are lots of ways to make important progress with a mouse or fish model, or other animal model.
- You can see if the animal gets port-wine birthmarks or other vascular changes. I can tell you that yes, indeed they do.
- You can look at the cells to understand how and why they are changed when there is an R183Q mutation.
- You can test drugs to see if they can reverse these changes—and therefore develop clinical trials.

The goal is to cure the mouse!—cure the fish!—to figure out how to help people.





REPORT: ROOTS TO RESEARCH

ON THE ROAD AGAIN



Early in the history of the SWF, it was realized that part of the mission would be to carry the word about the Foundation, and about Sturge-Weber syndrome, to the people who need to have medical and professional knowledge about SWS as well as to the families and individuals living and coping with it.

Every medical specialty in the US has an organization of physicians and allied clinicians who have the task of educating their members, advocating for their profession and advancing the science. The first job for the SWF was to identify who these organizations were and to gain entry into their membership lists, either as an affiliate or as a professional “guest”.



Through diligence and persistence, the SWF, through its officers and staff (small as it was with no large corporate structure), was able to carry its name and reputation into the mix of organizations who are the chief allies the SWF enjoys today. The list is like a virtual alphabet soup of organizations from the ABC (American Brain Coalition) down to the SfN (Society for Neuroscience). Many of the organizations partner with other groups for mutual benefit and often members belong to

several associations. A member of the AAD (American Academy of Dermatology) may also belong to the CSD (Coalitions for Skin Diseases) and the SID (Society for Investigative Dermatology).

The governmental agencies also wield enormous influence and power and the SWF needs to be conversant with their workings – like the NIH (National Institutes of Health), NINDS (National Institute for Neurological Disorders and Stroke), NIAMS (National Institute of Arthritis, Musculoskeletal and Skin) – all powerful friends. It is all focused on learning, networking, spreading the word and interacting with like-minded professionals and advocates.

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REPORT: ROOTS TO RESEARCH



ON THE ROAD AGAIN

(continued from page 17)

It takes time and money. Not just the registration fees, but the travel, airline, hotel expenses. Not to mention the personal energy expended in keeping the name and presence of the SWF before any number of audiences of medical, scientific and non-medical professionals.

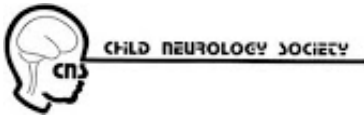
When you see or hear that one or two of the SWF staff members went to a meeting of the XYZ Association, you can be sure they were working for all of us.

ROAD UPDATES 2018

This Fall, the SWF attended the following annual meetings to distribute patient education materials, foster new clinical trials and engage with physicians, nurses, and industry attendees. These meetings are key opportunities to drive collaboration and create awareness to young clinicians and researchers. They CREATE the IMPACT for all of us.

CHILD NEUROLOGY SOCIETY (CNS) Chicago, IL

The SWF exhibited at the annual CNS meeting displaying new patient and healthcare education materials. The SWF Infographic piece provides highlights and key statistics on how the SWF creates impact for all we serve. The



Mission booklet details what we do and why we do each respective program or project. Brian Fisher, VP of Operations and Corporate Partnerships met with several key industry partners to discuss potential clinical trials and to engage the interest of other industry exhibitors. Karen Ball, President and CEO and Brian Fisher also had a

planning sessions with Jeffrey Loeb, MD, PhD, Chief Clinical Strategist, SWF Board Chairman, Kris Sadens and his wife, Kellie. They also hosted a luncheon meeting to discuss SWF activities with Anna Pinto, MD, PhD.

PEDIATRIC DERMATOLOGY RESEARCH ASSOCIATION (PeDRA), Denver, CO

This meeting was attended by Brian Fisher and Karen Ball to support Kaelin Ball who was invited to speak by Anna Buckner, MD conference chair; at the opening session on patient advocates. They also participated in the Port Wine Birthmark breakout sessions to collaborate on patient participation in new studies, genetic studies and to foster dermatology research studies. This burgeoning organization has made amazing strides since its inception and had 150 attendees this year!



PeDRA Conference; Kaelin Ball gives presentation





REPORT: ROOTS TO RESEARCH

ROAD UPDATES 2018

AMERICAN ACADEMY OF OPHTHALMOLOGY (AAO)

Chicago, IL

The SWF was invited to participate at the annual meeting's Informational Poster section. The latest poster was a work of art by our very own Susan Finnell, Director of Marketing. Brian Fisher and Karen Ball met with Clinical Care Network (CCN) ophthalmologists to discuss new projects and learn their needs at the local level. Industry partners were also engaged to discuss new clinical trial and marketing opportunities.



REPORT: 2018 CCN CONFERENCE



In September, the Sturge-Weber Foundation together with our host Clinical Care Network (CCN), Nemours Alfred I. duPont Hospital for Children, held a very successful conference. There were 20 CCNs out of 28 represented.

The conference cultivated discussion about where SWF is in areas like neurology, dermatology and ophthalmology, imaging, genetics/tissue banking and data integration.

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

REPORT: ROOTS TO RESEARCH

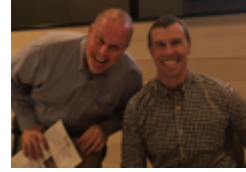


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The group discussed the next steps - trials, studies and additional research. Priority lists were made in each area. Protocols and transitions into adulthood was also discussed.

Insight from local families was a huge benefit and shed light on areas not previously explored. Dr. Falchek, Chief of Neurology was kind enough to give us the opportunity to tour the well-known garden area and see the beautiful mansion near the hospital. This gave us an opportunity to get a close up view of the grounds for the upcoming conference in July 2019.

The take away from this great conference were many new ideas, a great plan going forward for 2019 and renewed excitement from being together as a team.



*L-R: Dr. Pevsner;
Brian Fisher & Dr.
Burkhardt; Dr. Loeb;
Nemours Garden;
Sub-group at the
conference led by
Dr. Loeb.*



COLLABORATION

Two or more people working together towards shared goals.



BETTER TOGETHER

Catching Up With the
SWF Family



From Sherrie Harleman

My son Jesse who is 13, almost 14 years old and has type one SWS. He has never been able to tie his shoes, snap a button, or zip a zipper. The little things we all take for granted. Today Jesse did the zipper on his hoodie!!!! He was so proud of himself. I am over the moon proud of him!! For most parents, this is a mild accomplishment but for me, it means the world!!!

Design University. The Chief Scout's Award is the highest award which can be achieved at the Scout level in Scouts Canada (Same as US Eagle Scout). It was inaugurated in 1973 by then Governor General of Canada, Roland Michener. The award was created to provide recognition for Scouts when the Scout program was divided into the Scout and Venturer sections.



Connor and Caitlin Lawrence-Lovelace

From Leslie Greene

This is Connor with Caitlin Lawrence-Lovelace, receiving the Chief Scout Award. She is the grand daughter of Roland Michener, and my very best friend from Ontario college of Art &

From Julie Luby

(Allie Shepard's Stepmother)

At the end of August 2018, Allie Shepard graduated from Nursing School at Eagle Gate College in Salt Lake City, UT with a Bachelor of Science in Nursing. This is her second Bachelors degree (she also has a Bachelors degree in Health Education & Promotion from the University of Utah).

A month after she graduated, she successfully passed her NCLEX exam (National Council Licensure Examination) so she is now licensed as a registered nurse (RN).



It was a busy time for Allie--during the last four years, she worked hard to financially support herself while going to school, and was married in April 2017. All of her family members are so proud of all of her hard work and so happy that she can now pursue her dream of being a nurse.



From Jack Breeden

I wanted to thank Julia and Brian for their help and valuable references.

We contacted Dr. Geronemus and got an appointment within ten days. Thus 13 days later, we were in his office, he examined Cynthia, and removed the large growth within an hour. Then, yesterday he started the laser treatment to remove the coloring of the total birthmark.

Needless to say, Cynthia (and her RN sister, who works for a local dermatologist) are very pleased with the initial results. Cynthia has never smiled so much as when she looked in the mirror after the surgery. Donnie Hood, 35 and living in Canada will be journeying to Mallorca, a Spanish island to raise

DONNIE HOOD TAKES ON THE MALLORCA 312



Donnie Hood, 35 and living in Canada will be journeying to Mallorca, a Spanish island to raise awareness of Sturge-Weber syndrome and the Sturge-Weber Foundation.

Donnie made a decision at an early age to do everything he could to live as healthy as possible. exercise, and in particular, cycling, has been a big part of that. Cycling helped him to understand that having a medical condition like SWS should not define him, but instead, take every day as a blessing and always do his best to improve himself.

The Sturge-Weber Foundation was a huge step in helping Donnie progress with his life. He and his family found hope. The Foundation helped with medical issues and connected him to many people who have been a great support. With this on his heart, he wants to give something back.

Through the Mallorca 312, Donnie wants to set a meaningful personal challenge and test himself in one of Europe's longest and toughest amateur sporting events. The race goes through the beautiful but grueling mountain range of Mallorca.

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DONNIE TAKES ON MALLORCA

(continued from page 21)

With this race he will raise awareness of Sturge-Weber syndrome and the Foundation, and shine a light on the physical, psychological, emotional and social challenges that children and adults with SWS must endure, as well as its affects families on a daily basis.

Please join Donnie and us in this amazing journey. We will keep you updated on Donnie's progress and his incredible IMPACT!

BABY, IT'S COLD OUTSIDE!

Now available at the SWF Online store - Webster's Winter Cap and Scarf! Keep Webster warm this winter season as he travels with you!

Cap and scarf come as a set for \$11.00 (price includes shipping).



Keep those pictures coming and posted at the Webster, SWF Road Warrior Bear Facebook page!

SWF TASK FORCE UPDATE

(continued from page 8)

Advocacy Task Force has put together definitions and key words for advocating in your government. As Rare Disease Week comes up in February and other events it is important that you know the differences between each level of government. They are also working on PLANS to use when advocating. Stay tuned to the website for more information.

SWF TASK FORCE UPDATE (continued from page 6)

Surgery Task Force, (Candice Roberts, Chairperson) is working on the different kinds of surgeries and creating a glossary of terms. Once completed will look for resources and support groups to help manage as part of your Plan.

Before Their Time Task Force is focused on when we lose the ones we love too soon. We need your help if you are someone that would like to help with this Task Force we would love to hear from you.

Mental Health Programs for Younger Group Task Force, (Donnie Hood, Chairperson) has been focusing on identifying where to find appropriate and accurate resources related to mental health. They are researching websites that are interactive and easy to navigate. They want to include appropriate apps, games and even podcasts while also looking for interventions to improve mental health. They are also working with the Transition Group to develop a possible packet and or forum for both individuals and caregivers. All of this needs to be easy to navigate for the youth population so that we can be sure they have a place to go when the need arises.





Don't miss the greatest show on earth!
The 17th Annual Reunion of Champions
benefiting SWS/Birthmark Patients and Critical Research

Sponsorship details and online registration coming soon
at www.sturge-weber.org