The Sturge-Weber Foundation
12345 Jones Road, Suite 125
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FOR A REASON,
A SEASON,
OR A LIFETIME!

JUNE/JULY 2018

Branching Out

A PUBLICATION OF THE STURGE-WEBER FOUNDATION

MONTH OF AWARENESS IN REVIEW

SPOTLIGHT ON RESEARCH

SWF TASK FORCE

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The Sturge-Weber Foundation
MAGAZINE

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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

June/July 2018

IN THIS ISSUE . . .

On the Front Cover: Summer Andrews

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 CLINICAL CARE NETWORK

California:
- **UC-Irvine**
  Primary Contact: Kristen Kelly MD, Dermatology
- **UCSP Medical Center-San Francisco**
  Primary Contact: Ilona Frieden, MD, Dermatology
- **Rady Children's Hospital-San Diego**
  Primary Contact: Lawrence Eichenfield, MD, Dermatology
  Sheila Friedlander, MD, Dermatology

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

Illinois:
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- Akira Yoshi, MD, Neurology

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- Anna Pinto, MD, Neurology

Michigan:
- **Children's Hospital of Michigan-Detroit**
- **Primary Contact:** Csaba Juhasz, MD, Neurology Imaging
- U of Michigan Mott Children's Hospital-Ann Arbor
- **Primary Contact:** Jennifer Reeve, MD, Dermatology

Minnasota:
Mayo Clinic-Rochester
Primary Contact: Megha Tollefson, MD, Dermatology

North Carolina
- **UNC Children's Hospital-Chapel Hill**
  Primary Contact: Craig Burkhart, MD, Dermatology

New Jersey:
- **Northeast Regional Epilepsy Group-Hackensack**
  Primary Contact: Eric Segal, MD, Neurology

New York:
- **NYU Medical Center-NYC**
  Primary Contact: Daniel Miles, MD, Neurology

Ohio:
- **Cincinnati Children's Hospital-**
  Primary Contact: Adrienne M. Hammill, MD, Hemangiona and Vascular Malformation
- **Nationwide Children's Hospital-Columbus**
  Primary Contact: Warren Lo, MD, Neurology

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

Texas:
- **Cook Children's Medical Center-Fort Worth**
  Primary Contact: Saleem Malik, MD, Neurology
- **Dell Children's Medical Center-Austin**
  Primary Contact: Moise Levy, MD, Dermatology

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

Connecting with Karen

You Have Today

You have today. Just read a story about staying in the moment and not letting distractions shift your focus on what really matters and create glorious moments that stamp time and exude love. You know what I'm talking about…just one more email response…almost finished…the toast is burning! Off to your left, that small sad voice says, " OK mama, I'll wait". How many of those moments did I put them in second place for the good of the SWF or families with my two now adult children and TRUST ME you can't get back time. I try to let the guilt not reside, but slide!

I've had the pleasure of watching Crystal and Shad Elliers raise their amazing daughter, Carley, for the last 17 years and that recent story along with Carley's shocking 17th birthday…how'd time go by so fast…made me decide to share what we ALL know and TRY to do…be present…give yourself and your loved ones a PRESENT!

No better time to do so than summer and all the festivities the season brings! The Board of Directors will be meeting for our retreat and setting new strategic initiatives to bring you the best care and to drive crucial research investigations too. Please let me know your thoughts on what programs you'd like to see implemented, research conducted etc…we aim to please!

AMAZING Month of Awareness activities! Thank you to **Dr. Anna Pinto, Dr. Alex Levin and Dr. Kristen Kelly and staff** for sharing the awareness with the SWF T-shirt. The Speer and Werline families cranked it up a notch too with their fantastic local events and so many more of you writing posts thanks to Witney Arch's "noodging"! Each year gets better and better with more participants and funds being raised to further the mission and generate public awareness. Thank YOU!

This issue also has some great tips on Growing Golden. We're not leaving the pediatric cases behind just developing a library of tips from our adults who blazed a trail for you! The next SWF International Conference will be a terrific time of celebration, learning and uniting and between now and then I look forward to seeing y'all in Dupont, San Diego and Raleigh Durham.

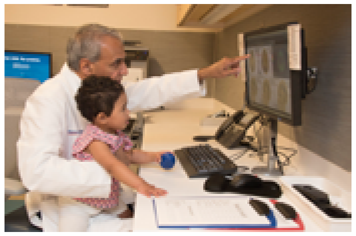
Have a wonderful summer and thank you for being with me on this awe filled journey with Sturge-Weber syndrome and Port Wine Birthmarks that so impact our lives and enrich us by their presence!

Faith, Hope and Love,

Karen L Ball

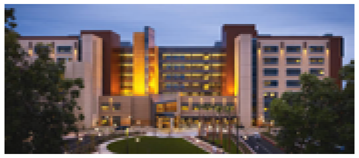
SWF Education Conferences

SWF Education Conferences are an excellent opportunity for patients, caregivers and professionals to come together as a community to educate, care and share. Each conference is located at one of SWF's Clinical Care Network centers. Check out the upcoming conferences planned across the country and visit with Karen Ball and SWF staff!



Nemours duPont Hospital for Children,
Wilimington, DE
Saturday, July 14, 8 AM - 2 PM
Dr. Harry Chugani, Pediatric Neurology
Dr. Michael Sperling, Transitions to Adult Care
Dr. Jonathan Salvin, Ophthalomology
Dr. Karen Wohlheiter, Psychiatry;
Carol Greene-Roethke, CRNP-APN
Nemours Clinical Dietician
Contact: Jennifer Hultber,
jennifer.hultber@nemours.org

Cook Children's Medical Center, Ft. Worth, TX
Hochberger Auditorium, 801 7th Ave.
Saturday, October 20, 8 AM - 12 PM
Dr. Dave Shahani, Neurology
Dr. Alan Norman, Ophthalmology
Dr. Heather Volkman, Dermatology
Dr. Kia Carter, Psychiatry
Contact: Amy Davis,
amy.davis@cookchildrens.org



Univeristy of California - Irvine, CA
Saturday, October 27, 8 AM - 2 PM
Discussions with Experts on research and treatments, ask questions about PWB, Glaucoma, Neurology and more.
Details coming soon on guest speakers and how to register.

Extending Our Reach

SWF is launching a new Patient Engagement Network (PEN) initiative. The PEN Task Force Teams will utilize volunteers and their unique talents. This is an opportunity to join forces with people of diverse experiences around the world. This united collaboration will create lasting bonds of friendship and further the Sturge-Weber Foundation Mission.

The SWF Task Force is a natural extension of the PEN, established in 2016. The Task Force consists of twelve teams that will focus on quality of life and medical treatment matters and foster respect for cultural differences.

Each team will consist of a chairperson and 3-5 members that are comprised of a patient, caregiver, and/or healthcare provider. All members will apply to be a part of the PEN Task Force and will be selected based on interest, knowledge, time and talent.

If you are interested in participating in the PEN Task Force and would like to review a list of teams and apply, visit the SWF website at www.sturge-weber.org/participate/.

If you have questions, contact Julia Terrell, jterrell@sturge-weber.org.

> ***"The PEN Task Force will extend our reach across the globe with vetted resources and manpower beyond our current capacity. More importantly, it will empower those who participate in the Task Force."***
>
> ***- Karen Ball***

We Are
Better
Together!

The 2018 TeamSWF Falmouth Road Race is Sunday, August 19 this year. This is the 9th year a team of runners has represented the Sturge-Weber Foundation in this renowned road race. Each year the team has grown, starting with only 5 runners the very first year. Over $595,000 has been raised by the runners to date. Once again, Pam McIntyre and Jessica Melo, two champions of the SWF and mothers of sons with SWS, will be leading and motivating the team of runners. We are so grateful for the continued enthusiasm and support of these two women!

Check out this amazing Honor Roll of Falmouth Road Race Warriors:

9 Years - Pam McIntyre, Jessica Melo
7 Years - Chris Marino
6 Years - Michelle O'Connor, Octavio Melo, Jonathan Desmaris
5 Years - Nancy Carpenter, Kate Sanders
4 Years - Erin Kelley, Brent Crouch, Mike Collins, Julie Brady, Woody Crouch
3 Years - Peter Sullivan, Carol DiPietro, Melanie Arena, Kim Auen, Mark Banks, Olivia Melo, Trish Bourne
2 Years - Lisa Ballantyne, Alicia Fannon, Kym Fischer, Kate Tiberio, Dan McIntyre, Courtney Mello
Newcomers - Kellie Sadens, Kate Schultz, Jen Pagliuca, Mark Fischer, Graham Ballantyne, Chris Wicken, Jason Cafarella, Jasmin Khamis, Will Carpenter, Josh Reed, Bianca Pyko, Ashley Reed, Paul Grew, Jameson Melo, Ben Bourne, Natalie Bourne

If you would like to sponsor one of these Road Race Warriors, go to the 2018 TeamSWF Falmouth Road Race page at: https://www.firstgiving.com/event/SWF/2018-TeamSWF-Falmouth-Road-Race

Sturge-Weber Connection

The Sturge-Weber Foundation Support Network and Discussion Community

Now there is another way to connect with others who live with Sturge-Weber syndrome and Port Wine Birthmark conditions. Introducing the Sturge-Weber Connection, an online global community by Inspire.

The Sturge-Weber Connection is very similar to Facebook, but provides a much more ***private and safe*** environment for discussion on sensitive details about SWS and PWB. Our community will have control over personal information and be able to archive it for future knowledge. Learn from medical experts about the latest research. Patient, caregiver and professional will be able to collaborate, share stories and discuss questions and concerns.
And the best part, it's totally FREE to you.

Ready to join? Let's get started then!
- Go to: ***https://www.inspire.com/groups/sturge-weber-connection/***
- Click on the button "Join This Community"
- Follow the basic instructions and register yourself.
- Once completed, you will receive a confirmation email.

That's it, you are all set and ready to CONNECT!

AMAZING AWARENESS MONTH!

During the SWF Month of Awareness, YOU stepped up to the plate, and YOU hit it out of the ball park!

What an amazing Month of Awareness for SWS from coast to coast! The staff and Board of Directors cannot thank you enough for your time, treasure and talant that was given to raise awareness.

Here are just a few highlights from awareness events held this year:

The Andrew's Family (Elizabeth and Summer) - televised awareness for SWS in Johnson City, TN on May 1st.

The Werline Warriors (James, Marla and Camila) - Chipotle Fundraiser on May 7th in San Antonio, TX raising $1079.

The Fit Revolution (Pam McIntyre) - an annual fitness event during MOA held in North Reading, MA on May 12th.

Myla's Mission Walk (Molly, Thomas and Myla Speers) - their first organized 5K walk in Greensburg, IN on May 19th raising over $12,000.

The Werline Warriors and CycleBar - a fundraising cycle class and raffle in San Antonio, TX on May 20th raising over $1500.

Jayden's Smile Bingo Fundraiser (Kathy Robbins) - an annual Bingo party during MOA held in Santa Ana, CA raising $4,400.

Trisha's Tots Dance Studio (Trish Bourne) - a raffle held at the dance studio's recital each year in Tewksbury, MA.

THIS IS JUST A SNAPSHOT of all the hard work and heartfelt support that was provided this year. The IMPACT continues, let's keep the ball rolling Team SWF!

GROWING GOLDEN PART 3: DERMATOLOGY

This issue highlights some common concerns about skin care. Craig Burkhart, MD, of the UNC Children's Hospital in Chapel Hill, NC, an SWF consultant, offers up-to-date information for adults who may not have had laser treatment recently.

Dermatology questions and concerns from adults. Thanks to Jillian Mills of Montreal for posing these questions and concerns.

For someone who hasn't had laser treatments in over 15 years, what might have changed in terms of length of time (duration) of treatment, recovery time, number of treatments before seeing results, etc? I remember back then thinking I was under the laser for hours.

The pulsed dye laser is the most common laser used to treat port wine stains today—just as it was 15 years ago. However, improvements in the technology have made the laser much faster and flexible than the earlier devices. Some things you may notice are (1) that the laser may be less painful as the cooling technology has improved; (2) the recovery time may be shorter as the "pulse profile" has improved; and (3) the session is much faster. Additionally, newer lasers are able to deliver higher energy to the skin than previous lasers, which may allow some previously resistant spots in the port wine stain to fade. Lastly, a larger variety of lasers are used to treat port wine stains today and your provider may be able to offer you alternatives to the pulsed dye laser that may work better for your skin.

If our skin gets dryer and thinner as we age, how will laser treatments affect it?

Your recovery time may be longer after each treatment as you get older—you may get more bruises that take longer to go away. Where you may have recovered within 1 week as a child, it will more likely take the full 2 weeks for your treatment spots to fade as you get older. You can ask your laser surgeon to modify your laser settings, however, to see if you still achieve the same results without extensive bruising. Protecting your skin from the sun during and between treatments is the best way to prevent drying and thinning of your skin as you age.

Is there a "right timing" for restarting treatments? Things seemed of be more or less stable for a few years but now there seem to be moles/blebs popping up almost overnight and my skin is itchier.

I would say the right timing is when you want to change something about your birthmark. If you are happy with your birthmark and it is not bothering you, you do not need to treat it. If you are developing blebs and moles, I would recommend treating those areas to prevent the blebs from getting larger or bleeding. If the skin over your port wine stain is itchy, I would have the area evaluated by a dermatologist to make sure there is not a primary skin disease causing the itch (like a dermatitis or fungus). If there is no skin disease causing the increased itch, then I would do a trial of lasers to see if that helps your symptoms. Do not do laser treatments when you have an active sunburn or tan.

Is it possible for conditions to have progressed too far to be dealt with by lasers?

Lasers only treat the top layer of your skin--they can treat color, texture, and small bumps on your skin. Skin lasers cannot change the overall shape of your face. If the contour of your face (overall shape of your face) is what concerns you, then surgery would be more appropriate than lasers.

What issues are important to discuss with my doctor that I might not have thought of?

Remember to let your doctor know about important events that are coming up in your life. You will want to schedule your treatments around these events—you don't want to be dealing with bruises during a wedding photo shoot. Also, having a visible birthmark can be very stressful! Let your provider know if you are becoming "stressed-out" or becoming depressed or anxious due to your birthmark or its treatments. Although your laser surgeon may not know a lot about mental health, he or she should be able to walk with you through the course of your laser treatments and help you find social supports and mental health resources within your community.

Beyond laser, what could be necessary like mole removal, tissue debulking etc?

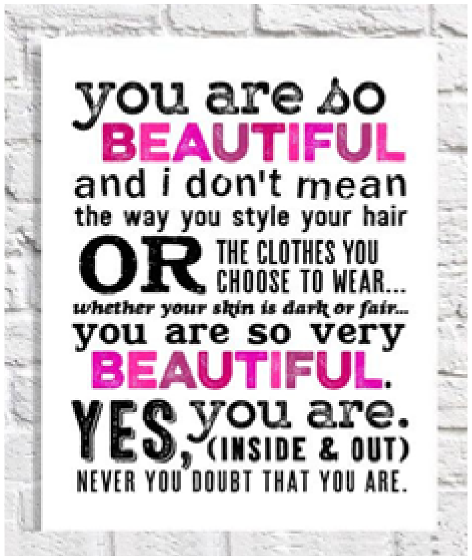
If you have changes to the overall shape of your face (contour change), that you would like to modify, surgery can be used in addition to lasers make these adjustments.

What sunscreens are recommended for extremely sensitive PWB skin? I try to limit time in the sun – with SPF #30 or higher – to no more than 5 minutes.

If your birthmark is extra sensitive, sunscreens based on physical blockers tend to be the least irritating. Physical blockers are titanium dioxide and zinc oxide. Creams and sunscreen sticks tend to be less irritating than gels and sprays. However, sometimes you have to experiment with several sunscreens before finding the brand and type that works best for you.

Would it be healthier for my skin not to use makeup?

Most makeups do not cause any bad effects on the skin. If you notice that your skin breaks out with acne bumps when you wear makeup, then look for brands with a label that states "non-comedogenic" or "do not cause acne." If you notice that your face becomes more itchy after wearing makeup, you may be allergic to certain makeup ingredients. If you have this symptom, a dermatologist or allergist can do patch testing to find out if you are allergic to makeup and what makeups are safe for you.



GROWING UP CAREGIVING

It is not the job of the sibling in any family to be the caregiver. The primary responsibility, as always, resides with the parents. But parents who create and foster an atmosphere, perhaps unspoken, of care and competence in their other children, lay the groundwork for the future so that siblings can move into a caregiving role if and when necessary. This can enhance the natural love and concern children have for their brother or sister.

But the relationship is not without certain complexities. How much responsibility do parents expect of child who may be only a few years older than the patient? Do they shield the siblings from the medical concerns and take care not to involve the sibling? Do they create a family atmosphere where the care of the patient is shared according to the ability and understanding (and aptitude) of the sibling? SWF families are intrinsically involved in these concerns.

The community and medical resources all special needs persons need are primary and essential components of care. But the relationships of brothers and sisters are constantly present. Families share their experiences.

Michele Barton tells us about her big brother Chuck Stewart.

Charles H. Stewart (Chuck) was born October 31, 1951. Chuck was born with the port wine stain. According to my mom, she knew something was not right with Chuck.

Chuck had seizures from the time he was a baby until a young boy. He went through all kinds of testing at Walter Reed Hospital and Ochsner Hospital in New Orleans. Ochsner's made the final diagnosis of Sturge-Weber Syndrome.

This diagnosis was made in the 50-60's timeframe. This was an unknown disease and no one knew how to treat it. In an attempt to control the seizures, Chuck was paralyzed. The doctors believed the paralysis was permanent, my mother did not. My mom is the reason that Chuck is walking, standing, sitting and talking today.

My mom went to every doctor, school board supervisors, anyone who would listen. Letting them know that Chuck would progress if given the opportunity. I don't believe she had an easy battle in the 60'-70s; but she didn't give up.

(continued from page 12)

My dad was in the service, so Chuck is known as a 'military brat'. He went to public schools in New Orleans, LA, Newport News, VA, the American School in Vicenza, Italy, public schools in Augusta, GA, Orlando, FL, Hattiesburg, MS, Lompoc and Sacramento, CA.

He went through special education and received a high school diploma. He has a driver's license, but currently doesn't drive. He worked in food service for 30 years at the University of Southern Mississippi, in Hattiesburg, MS. He retired after being diagnosed with cancer in 2005. Chuck survived the cancer. He is loving retirement. We lost our dad in 2009 and it was hard on Chuck, but he stepped up and did what he could to help my mom and me.

(Photos on page 12: Chuck and Michele in 1960. Chuck and mom Sophia 'Kitty' Stewart.)

Melanie Arena, mom of Evan and Blake shares this.

I love this idea to celebrate siblings as caregivers! I wrote a Facebook post for Awareness Month about how my 7-year-old son Blake is an incredibly important caregiver of his younger brother Evan.

Our family is so blessed to be surrounded by a tribe of amazing people who support and care for us in so many ways. However, Evan's number one caregiver, who often gets overlooked, is Evan's best big "brudder", Blake. Blake gets called upon to step up more often than most siblings. I have held him to higher standards because I was exhausted from the extra pressure of caring for Evan and I didn't have enough energy left to give. Blake is often quietly performing in the background while we celebrate every little milestone that Evan makes. Blake is Evan's best friend, closest role model, and biggest motivator.

This picture is from a year and a half ago, when Blake came to visit Evan in the rehab hospital after a few weeks apart. Evan was beginning to recover from seizures and hemiparesis, but he wasn't talking much and was still too weak to walk. Blake brought joy back to Evan. Evan began talking, playing and laughing more. Blake went to therapy with Evan, encouraged him, and took him on stroller rides. And two days later Evan was walking, well, toddling around on a playground trying to keep up with Blake.

These boys know how to push each other's buttons like siblings do, but there is no one that Evan looks up to more than Blake. Evan models Blake's example, and Blake can often be caught in the act of teaching Evan how to do something. Evan looks forward to time spent with his big brother more than anyone else. We don't tell Blake this enough, but we are so proud of the caring, smart, creative, hard-working, and independent little man that he is. And we are so lucky to have him supporting Evan all the way.

More Growing Up Caregiving Stories on page 14.

GROWING UP CAREGIVING

The Knoblach family of Arizona have four sons. Conni and Jerry have been with the SWF since shortly after Aaron was born in 2003.

Their two older boys and their youngest, have grown up learning how to navigate around having a sibling with a rare disease.

Oldest brother Andrew Knoblach is now in college and shares some experiences.

Things always seem a lot less rushed now that we have to slow down sometimes for Aaron and take more breaks. Also I'll never forget all the back and forth trips to the hospital when Arron had seizures.

I remember when I was really young being told how to watch for signs of a seizure by my dad and then, not 10 minutes later, I had to go and get him because I saw Aaron having a seizure.

On one family vacation I had to backpack Aaron across Europe until my knees almost gave out. Still worth it and a good work out.

Over time I just had to learn a little more patience when doing things with Aaron, whether it bewalking upstairs or trying to figure out what he is saying.

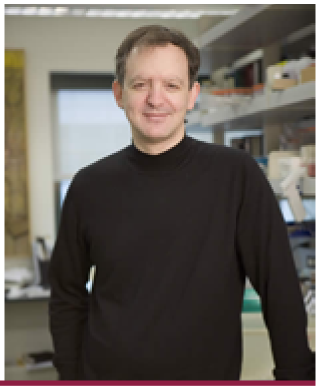
So I have learned patience, and how to imagine myself in someone else's shoes, and how not everyone has been given the same attributes and talents. But they have their own worth and can be happy and sometimes funny. As the saying goes "he's not heavy, he's my brother

We watched my parents deal with emergencies and daily chores and we came to understand that we could do this too.

Sometimes being a brother or sister is better than being a super hero!

SPOTLIGHT ON RESEARCH

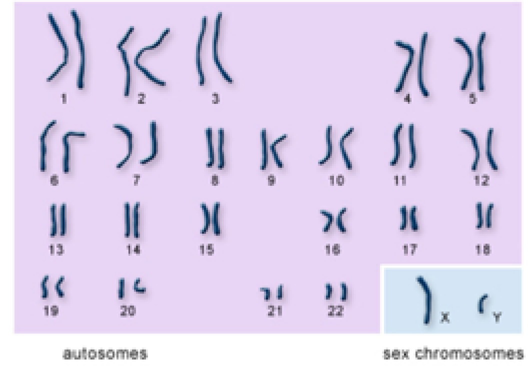
Dr. Jonathan Pevsner
SWF Chief Scientific Officer



What is a gene?
It turns out that we have over 3 billion base pairs of DNA, arranged in the famous double helix that Crick and Watson figured out in 1953. And all that DNA is organized into chromosomes—for girls it's chromosomes 1 through 22 plus two copies of X; for boys it's chromosomes 1 through 22 plus an X and a Y.

Let's look at chromosome 9. From end to end its length is 141,213,431 base pairs of DNA-that's 141 million of the building blocks (called A, C, T, G). And scattered along that chromosome we can find 2,253 genes.

So the gene is something made of DNA and is part of a chromosome. And it turns out we have 20,000 genes that make proteins (we'll talk about what a protein is another time). Genes are too small to see, but for over a hundred years we've known what they do. They can control eye color, or development of the limbs, or height, and all sorts of other features. They've been called the "blueprint of a house" and that's a nice metaphor. A house has walls, plumbing, stairs, and other parts that we see; similarly a cell has different features we can see including a transport system, a garbage disposal system, and walls. The DNA blueprint gets turned into proteins that let a cell do its job.



U.S. National Library of Medicine

What is GNAQ?
One of our genes is called GNAQ. It sits on chromosome 9, and it makes a protein called GNAQ (pronounced G alpha Q). NIH offers a web page that introduces it: https://ghr.nlm.nih.gov/gene/GNAQ .

To view a DNA sequence from an NIH website, go to https://www.ncbi.nlm.nih.gov/nuccore/NM_002072.4. There are 6,343 bases of DNA (using the alphabet "G, A, T, C").

(continued on page 16)

SPOTLIGHT ON RESEARCH

Dr. Jonathan Pevsner
SWF Chief Scientific Officer

(continued from page 15)

Why does it matter?
In Sturge-Weber syndrome and in port-wine birthmarks, just one of those bases changes.

The change is called R183Q.

Q: When does it happen?
A: Before birth.

Q: Does it happen in mom's egg or dad's sperm?
A: No, it happens by chance during development of the fetus.

Q:Where does it happen?
A: Mostly in endothelial cells.

Q:Why does it happen?
A: Mutations happen very, very often. They're part of life.

Q:What does it do?
A: It turns on the GNAQ protein--too much.

Q: How does it relate to cancer?
A:The same R183Q mutation can cause melanoma of the eye (uveal melanoma): it happens at a different time of life (not during early development), in a different cell type (melanocytes not endothelial cells), and people with SWS or port-wine birthmarks are not at increased risk for getting that cancer.

Let's cover some of these questions in other posts—for example I'd like to introduce you to the GNAQ protein, explain what it means for it to be turned on, and tell you how researchers are trying to figure out how to turn it back off.

Conclusions: the bigger picture
There are many ways to try to help someone with Sturge-Weber syndrome. Doctors, nurses, therapists of various sorts can all help understand the condition and its complications including the most effective treatments. Those of us involved in research are focused on understanding the underlying causes. Much of this happens at the molecular level, and dealing with genes and proteins and cells may seem abstract because we don't see them in our everyday lives. In this post I introduced the concept of the gene and mentioned GNAQ, the gene that harbors a mutation that causes Sturge-Weber syndrome and port-wine birthmarks.

I hope this gives you an initial background so we can talk about the many ways the research community can solve the puzzle of what causes the brain, eye, skin, and other complications of Sturge-Weber syndrome from seizures to birthmarks to headaches.

Here's what we can do, knowing that GNAQ mutations cause SWS:

- Knowing the gene we can study how it works inside a cell, and what the consequences are of the mutation
- We can study how it works in many other organisms (many of which are far easier to study than in humans)
- We can create the mutation that causes SWS in model organisms (such as mouse and fish), see the effects, try to fix them, and take what we've learned back to patients with SWS
- Knowing that the same mutation in the same gene can cause uveal melanoma we can see what strategies the cancer community has taken to solve the effect of mutated GNAQ.

All of this is meant to help us get close to figuring out treatments that can help treat the root causes of SWS.

BETTER TOGETHER

Catching Up With the SWF Family

Remembering Loved Ones

Our heartfelt condolences go out to their families on the loss of these members of our Sturge-Weber community.

Lois Potts, grandmother of Dalton Potts-Lesage, passed away in March 2018. in Kansas. Her daughter, Lori Kroboth, is Dalton's mother. They have been SWF members since 1998.

Tom Spartis passed away on April 27, 2018 in Pittsburg, PA. Tom has been a member of SWF since 2013. He was 67 years old.

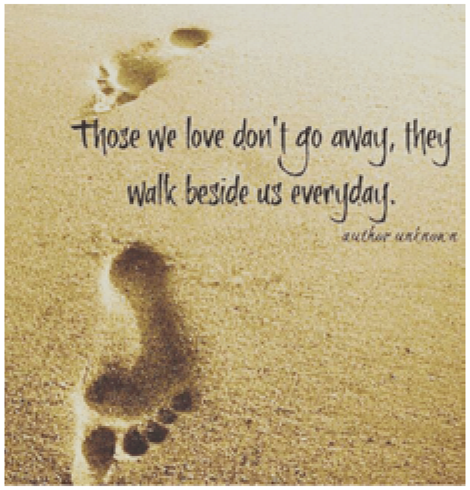
Frances Walsh, grandmother of David Brown, passed away on March 18, 2018. She was a faithful supporter of David's family and the SWF since 1993.

Chad Layman of Florida passed away on May 4, 2018. He was 47 years old and lived with his parents Fran and Larry. Chad was an Eagle Scout and an active community member.

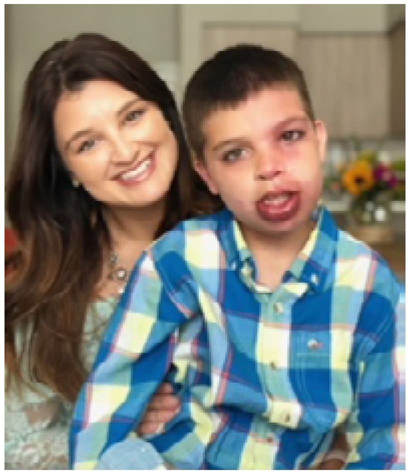
Paul Siegel passed away on May 17, 2018 at his home in MD. Paul and his late mother Lynne were active SWF members for several years. Paul was 36. They had attended several SWF conferences and local activities. Paul was a proud college graduate who pursued his degree for 15 years. He credited his mom with teaching him how to be an advocate for SWS.

Thank you to the Kroboth/Potts, Spartis, Siegel and the Walsh/Brown families for their memorial requests to the SWF. Such generous donations can sponsor another person to receive a SWF Conference scholarship or plant a seed for research in seed grant funding.

We are always grateful when families, in their time of sorrow, include requests for memorial donations to the SWF in their public notices. Your generosity helps continue the legacy of caring that benefited your loved one.



My Favorite Caregiver



Nikita and Lathan Wiggins

During the Month of Awareness we received nominations for favorite caregiver. This is the story of Nikita Wiggins, submitted by her mother. This story portrays the love and support that ALL caregivers provide selflessly.

My daughter, Nikita Wiggins, her son's, Lathan, favorite caregiver.

She is his mom and of course does all the normal "mom" things, however, I have been on this journey with her for 11 years and watched since the beginning this young mom ensure he had all the therapists needed at a young age to help him develop into the strong young man he is today. She hasn't missed very many of the doctor, dentist, optometrist, dermatoligist treatments and all the other important ARD meetings at school.

She has him involved in children's church choir, soccer, Boy Scouts, baseball and Special Olympics all while she works, going to school and caring for a newborn.

She has taught him about friendliness, respect, manners, helping others and standing on his own. It's amazing to watch Lathan now as a leader of his own which is in part to Nikita's leadership.

She has in turn been very instrumental in sharing her story with others that have children with special needs and helping them along the way navigate this very special journey.

This is the same mom at 20, heartbroken when Lathan was diagnosed and had his first seizure at 3 months old, that said, "I can't do this". She HAS done it and a fantastic job of providing Lathan with a great quality of life!

Love,
Lathan's Granny
Deborah Legate

Reaching the Top of the Scouting Trail

Congratulations to Joshua Fondren for reaching the level of Eagle Scout. Joshua, son to Sharon and Mark Fondren in Hollywood, received the highest level in scouting at an Eagle Court of Honor on Saturday, June 2, 2018 at St. John Francis Regis Church.

> **Caregiving often calls us to lean into love we didn't know possible.**

A Very Special Dedication

Gabriel Dalton, son of Sam and Mary, of Prince Edward Island, Canada, were more than just proud when they received word of Gabriel (Gabe) being honored through the dedication of the 2017-2018 school yearbook at Greenfield Elementary.

"This year we are dedicating this yearbook to a wonderful Greenfield student who will be leaving this year to go to Summerside Intermediate School. Gabe Dalton has been a student of Greenfield for seven years and he has been a great example of a Greenfield Gator - working hard at learning, always making many friends and helping out whenever he can. We will miss him at Greenfield, but as Gabe knows - once a Gator always a Gator!"

Father's Day Around the World

Father's Day may be a special day only in the USA, but for two SWF dads, it is every day in their part of the world. In addition to being the "paterfamilias" in their families, they have added their own special talents and concerns to the care of their children and the willingness to reach out.

Sam Dalton lives in Summerside, Prince Edward Island, Canada. Since his son Gabriel was born and their family joined the SWF in 2006, Sam has been a dedicated correspondent and reporter. Very soon he volunteered to contact other dads and share his empathy and experience with them.

One of the first international dads he linked with is Alekander (Sasa) Vujacic in Montenegro, Serbia, in Europe (the former Yugoslavia, on the Adriatic Sea). His daughter, Elena, is 12, the same age as Gabriel.

The two dads soon became the "go-to" persons we sought when other concerned parents, especially in countries with less access to care, contacted the SWF for assistance. They have never met, but that has not prevented a firm and friendly bond that has included contacting other families in the Middle East and Europe.

New Love, New Life!

Congratulations to Nick Bourne and his lovely wife, Rachel, married on December 9, 2017. May the rest of your days be filled with love and laughter together!

CALLING ALL WARRIOR MAMAS!

Karen Ball

I am closing in on getting the draft copy "Warrior Mama" to the editor! I would so like to include your wisdom as well!

Email me (kball@sturge-weber.org) a short aka, no more than 3-4 prapgraphs alonge with a photo of what you have done to be an advocate for your child OR if you were diagnosed with SWS or a Birthmark, too. It can relate to your tenacity, courage, resilience, vulnerability, effectibe communication shills, fellowship and more!

Also, what would you tell your younger self about become a Warrior Mama? You know this war we wage takes ALL kinds of caregivers; those of us on the frontlines, those of you active on the local scene, and those who exude patient love and understanding. let's tell the world all about it!

Please submit by July 15, 2018!

TEXAS
Health and Human Services

CONGRATULATIONS TO JORDAN WOOD

Jordan Wood received his Texas Nurse Aide certification from the Texas Health and Human Services Commission. Congratulations Jordan!

Shout Out to SWF Families

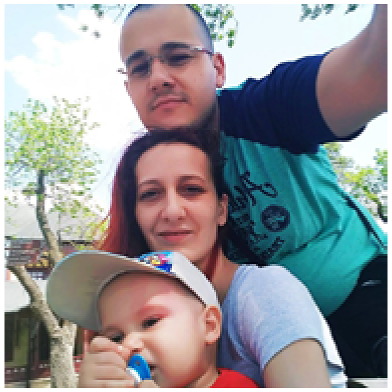
Sydney Willard's Aunt Kristen made chocolate covered pretzels that were sold during Month of Awareness! Thanks Aunt Kristen, please send some to Houston!

To Serbia With Love

Meet the Mijalovic's from Serbia. Luka is 1 year, 4 months old. He was diagnosed with SWS at 9 months old when he had a seizure for 1 hour. Since then, he has had 5 other seizure attacks and is on several medications to control them. Fortunately, Luka does not suffer from glaucoma.

Mina, his mother, says Luka is a great little warrior. She is amazed how strong the SWS children are. Luka is happy, and bring them so much joy. He loves people and sends kisses to everyone and waves vigorously!

The family is grateful there is a foundation that helps and educates them about SWS. They have learned to never give up and to stay strong!



It's hard to believe that the 2017-2018 fiscal year for SWF is coming to a close on June 30th. What an amazing year it has been.

It was kicked off with the 2017 International Family Conference in Cincinnati, OH. The SWF Clinical Care Network (CCN) was established, 25 centers and growing. These centers came together for the first SWF CCN Conference in New Orleans, LA on October 1-3. Members and newly diagnosed families have been connected with specialists throughout the country to get the best care, second opinions and much needed guidance through the CCN.

The Foundation was able to award another research fellow with a $50,000 research grant. Colette Bichsel, PhD of Boston Children's Hospital received 2017 Lisa's Sturge-Weber Foundation Research Fellowship Award.

In February 2018, the Foundation was able to raise funding for another fellowship award at the Reunion of Champions held at the annual Amerian Academy of Dermatology Conference. The Richard Felten Fellowship was created which will provide funding for fellows in the dermatological profession.

SWF also launched a new social media platform for its members to have a more private and secure environment to share their experiences, ask questions and obtain more information on their needs as a patient and caregiver. This will increase our level of community and collaboration with members and healthcare providers.

The printed materials SWF provides to newly diagnosed patients and families was updated and printed. This material provides a foundation of information on SWS and PWB that will guide families through their personal journey.

Webster the Road Warrior Bear program was implemented and is making a positive impact in awareness. Through our Share a Bear campaign in December 2017, we were able to touch the lives of many of our members with an expression of hope and connection, reminding them they are not alone!

So much more has been accomplished that isn't mentioned here. None of it could be done without the continued support we receive from SWS families, friends of families, fundraisers, corporate sponsorships and other foundations. Check out Dr. Jeffrey Loeb's report on the SWF Clinical Care Network.

THE STURGE-WEBER FOUNDATION
CLINICAL CARE NETWORK

Dr. Jeffrey Loeb
Chief Clinicial Strategist

The Clinical Care Network (CCN) has come together and made amazing strides in building a team-based structure for advancing our understanding of SWS as a means of improving the lives of those with SWS and their families.

A highlight was the combined CCN and Patient Engagement Network (PEN) meeting in New Orleans, LA on October 1-3, 2017. The major goal of this meeting was to identify important unmet needs across the entire specturm of SWS including skin, eye, brain, imaging, tissue, research and most importantly, quality of life.

It was a monumental effort that resulted ina monumental publication tat is now in press in *Pediactric Neurology.* The paper defines important clinical and research priorities which suggests important medical interventions. Equally important, the meeting and publications brought together groups from all over the CCN to not only provide a snapshot of where we are, but to define where we need to go and develop actionable items to move things forward.

SUMMER FUN AND TIPS FROM WEBSTER!

Summer Trips With Webster

With Summer vacation upon us, many of you will be traveling or planning a "Staycation". Don't forget to take Webster along! Take pictures with Webster along your Summer journey and post them the the SWF Webster the Road Warrior Bear Facebook page!

Let's see how many different places Webster will visit, how many miles he will travel, and how many new people he will meet along the way spreading awareness for SWS!

Webster's Summer Sun Tips

Check with your dermatologist, but these are sun-sensible tips from our experienced families.

Sun block/screen is a must. You can get them with SPF 70 – and don't be shy about applying them often. Especially in humid weather when we perspire. A pharmacist can suggest sunscreens that are suitable for babies whose skin is tender.

Keep hydrated – drink water – or Gatorade or similar product that replenishes electrolytes.

Location is everything – it you live in a hot, dry climate like South Texas, watch for signs of sunburn, heat exhaustion or heat stoke in other family members who may be doing outside activities like garden work. Learn the proper first aid for these conditions.

Even in New England the sun and temp get high enough to create sunburn. Being on a beach or by a pool can magnify the effects of the sun – cover up - there are hats and shirts that have sunblock properties. Wear good sunglasses with UV protection. And loose long sleeves on shirts. And don't forget your feet – the instep of your foot can get a painful sunburn if not covered. Watching a town parade is great fun and sometimes you forget that the sun is watching down on the parade too. Use a fancy umbrella to add to the fun.