

DEDICATED TO THE LIFE AND CARE FOR PEOPLE WITH STURGE-WEBER
SYNDROME AND PORT-WINE BIRTHMARK CONDITIONS

SPRING 2023



Branching Out

35
YEARS UNITED

FOR 35 YEARS WE'VE
BEEN HERE FOR YOU!
A reason, a season, or a lifetime.

THE SCIENCE AND RESEARCH ISSUE

CELEBRATING

10
ANNIVERSARY

GNAQ SOMATIC MUTATION
DISCOVERED
MAY 23, 2013

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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

SPRING 2023

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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.



Founder | CEO

Connect with Karen

It's About the Journey!

I have had the great honor to be Bev and Stan Fisher's daughter for 65 years. Some of their 93 years after I came into their lives I'm sure were longer than others with lil ole "Kar" "Kedder"! While other years, their hearts would be bursting out of their chests at what has been accomplished with my family and The SWF.

The CEO of Korn Ferry said Neuroscientists call the moments when your memory is crystal clear a "flashbulb memory". We remember them because of the emotional intensity. It's crystal clear where you were when the planes hit the towers in September. The place you were when you received the SWS diagnosis. The day you heard they found the gene for SWS, or the day you heard your mama and daddy died.

However, when we look back, we can still feel with clarity those celebrations and dark days when we celebrated our accomplishments or faced our fears of the unknown.

Now, time has moved on and it's years later. A lot can happen in that amount of time. For me, I graduated college, sang for a year around the world, taught school, got married, had a daughter, started the SWF, had my son, battled an arcane school system for my son and moved 4 times in the interim.

"History gives us an anchor, so we remember our mistakes to never repeat and what to never do or say again."

That raises the question: What about your own growth and learning during the ensuing years?

The truth is, no matter how much we'd like to put many years behind us, a larger danger is in forgetting to remember. As crazy as it may seem, out of the most traumatic events can come the most tremendous gifts: forest fires generate new growth, a SWS diagnosis becomes manageable, a divorce brings hope and peace, and even tragically Towers come down and unite a nation.

I remember the importance of others in your journey. Usually while I'm in the shower or outside when the butterflies or geese fly overhead, I now know they are reminders to remember those gone too soon from us. They are celebrations of life and the journey we shared along the way. Aaron Novak is my geese memory. My Gramma Lou turned me on to butterflies. Kimberly Slater is now my sunflower celebration. My mind is like a steel trap or maybe an elephant who never forgets...hmmm!

The courage, tenacity, and sacrifices of so many remind me there are warriors battling on many fronts every day to defend us, educate us, heal us, rescue us, and love us. I still remember the day Dr. Jonathan Pevsner told

Continued on page 16 >>>

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COVER STORY: 10 Years After GNAQ R183Q Discovery

MATT SHIRLEY, PHD

SWF Chief Science Officer

Contributor to “Sturge-Weber syndrome and Port-Wine Stains Caused by Somatic Mutation in GNAQ R183Q”, May 23, 2013

As we celebrate the **10th anniversary of the discovery of the GNAQ R183Q mutation that causes Sturge-Weber Syndrome (SWS)**, it's important to recognize the significance of this discovery and the progress that has been made in understanding and treating this rare disease. The discovery of the GNAQ R183Q mutation was a critical turning point in the study of SWS, and it has paved the way for new research and treatments that have the potential to greatly improve the lives of those living with this condition.

“...it (GNAQ R183Q discovery) has paved the way for new research and treatments that have the potential to greatly improve the lives of those living with this condition.”

In my first *Roots in Research* article as CSO I shared my experience working on the discovery of the GNAQ R183Q mutation in 2012. We relied on generous tissue donations of skin and brain, along with a modest research grant from the Sturge-Weber Foundation, to read out the entire DNA sequence from affected and unaffected tissue biopsies. With the help of a newly developed method from cancer genomics, they were able to pinpoint a single mutation in the GNAQ

R183Q gene that seemed to cause SWS. This groundbreaking discovery has since been confirmed by many researchers around the world, and we've learned more about the specific cells in blood vessels that harbor the mutation.

Since the discovery of the GNAQ R183Q mutation, significant progress has been made in understanding the disease biology of SWS, and researchers have been hard at work developing new tools to study and treat this condition. The Sturge-Weber Foundation has provided grant funding for several critical areas of research, and SWF-funded researchers are using mice and zebrafish engineered with the GNAQ R183Q mutation to develop systems to study SWS outside of the human body. This type of work is critical to enable a better understanding of the disease biology and for testing the safety and efficacy of new pharmaceuticals and therapeutics.

The Sturge-Weber International Research Conference held in July 2022 was a critical gathering of influential researchers and clinicians from around the world who shared their new findings and upcoming research on SWS. This conference helped connect the dots between the laboratory scientists and the clinical specialists and sparked new paths to understanding ways to improve the lives of people living with SWS.

As we look toward the next 10 years, research on Sturge-Weber syndrome is likely to continue to focus on understanding the disease biology and developing new treatments. Building on the discovery of the GNAQ R183Q mutation as the cause of



Q&A with Doug

Thoughts on the GNAQ Mutation

DOUGLAS A MARCHUK, PHD

James B Duke Professor

Department of Molecular Genetics and Microbiology

Duke University School of Medicine

Where were you when the gene was discovered?

Such a major discovery is not quite a single event that occurs on a single day. My lab was part of the team participated in the discovery of the somatic mutation causing SWS. After Matt Shirley and Jonathan Pevsner identified the GNAQ R183Q mutation in three patient samples, my lab performed a lot of the validation work (finding the same mutation in a larger bank of affected tissues from SWS patients) and then the functional research—biochemical studies to prove that the mutation caused activation of the GalphaQ protein (the protein encoded by the GNAQ R183Q gene). *So where were we?* We were on the front lines in the lab participating in the research.

What excited you most about the gene discovery?

I was excited because for decades, the underlying cause of SWS was unknown, and for the first time we finally knew the cause. For a while, until we finished all the experiments and collected all the data, and then submitted our paper to be published, we were among a small group of people that were the only ones in the world who knew!

Did it change your research?

It changed our research from trying to discover the cause of SWS to trying to understand how this mutation causes the vascular malformations. So we pivoted from discovery research to hypothesis testing research—that is, testing ideas about how the mutation affected the blood vessels.

Ten years later what is different today?

Ten years later we (the SWS research community) have multiple cellular and

mouse models to study the events leading to vascular malformation development in SWS. Importantly, these same models can be used to test therapies. We are well poised to make significant progress in the next 5–10 years.

Treatment and cure what's next in your opinion?

I believe we will be testing a treatment for patients in a clinical trial within 5 years. Possibly earlier, possibly a bit later. The hope is that the first or one of the first drugs tested in a clinical trial shows positive results for the patients. Until a drug is tested in a controlled clinical trial and it shows positive results, no drug will be approved for general use. But I believe that soon we will find a treatment that has a strong impact on SWS patients. As for a cure, a true cure, is a very high bar to set. It is difficult for me to think of very many diseases for which we have a true cure. And since the somatic mutation that causes SWS happens during development (in the womb), the effects of the mutation are already present by the time the first diagnosis of SWS is made in an infant or child. So it isn't going to be very easy to fully reverse the effects. But I think we will have very good treatments soon.

If you could tell our patients and caregivers to have hope for the future what would you say?

I have high hopes for a clinical trial soon. I would tell patients and families to get ready to enroll in a trial for an experimental therapy. We cannot be sure that the first drug that is tested will work, or if it does, work as well as we had hoped. But please be willing to participate for the good of your child/family and the good of future patients and families. We will get there if we work together.



Q&A with Joyce

Thoughts on the GNAQ Mutation

JOYCE BISCOFF, PHD

Professor of Surgery at Harvard Medical School with a primary appointment in the Vascular Biology Program and Department of Surgery at Boston Children's Hospital

Where were you when the gene was discovered?

To be honest, I don't remember exactly but I remember seeing the NEJM article and being excited about it!

What excited you most about the gene discovery?

What excited me most is that the mutation provides a launch point for basic science.

Did it change your research?

Yes, we began working with Arin Greene and Anna Pinto to determine which cells in the capillary malformation have the GNAQ R183Q mutation. It was a brand new project in my lab and now it's a major focus.

10 years later what is different today?

We've learned a lot about how the GNAQ R183Q mutation alters endothelial cell behavior, but we have so much more to learn.

If you could tell our patients and caregivers to have hope for the future what would you say?

Basic science is painstakingly slow but there can be sudden leaps forward when labs are working together and sharing results, which we all do thanks to The Sturge-Weber Foundation!



A Note from Jeff

Thoughts on the GNAQ Mutation

JEFF LOEB, MD, PHD

The SWF's Chief Scientific Strategist

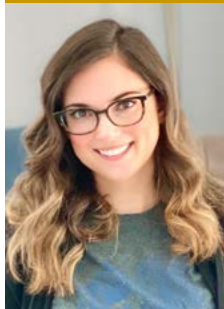
I had not yet gotten involved with SWS when the GNAQ R183Q mutation was discovered, however, having this discovery gives us enormous opportunities to translate this discovery into new treatments. That's my number one job at the Foundation! Knowing the gene mutation is a critical first step and we need to keep working forward to explain how the mutation leads to changes in the brain, eye, and skin.

We also have to work backwards from understanding the clinical symptoms that

are of most concern to those with SWS so that we can meet in the middle with new ideas and new treatments. My other priority is collecting accurate clinical information from SWS patients all over the country into the **BVMC database** to identify from past experience what treatments might be working and other that may not be helpful. I am confident that this database will successfully work backwards toward the gene discovery and we will have new treatment approaches and clinical trials for these very soon!

Learn more by clicking the link above.





Sturge-Weber Syndrome Research Poses a Unique Challenge

COURTNEY SHIRLEY PH.D.
SWF Science Director

Sturge-Weber syndrome research poses a unique challenge. Despite the fact that researchers have identified the cause of SWS, there is currently no known cure and treatment options are limited to managing symptoms. SWS is caused by a genetic mutation in the GNAQ R183Q gene, which leads to abnormal blood vessel development in the brain and skin. The GNAQ R183Q mutation happens sometime during embryonic development and is not in every cell. Animal models, particularly mice and zebrafish, have become invaluable tools in rare disease research, including SWS, offering insights into disease mechanisms, potential treatments, and drug development.

Animal models, particularly mice and zebrafish, have played a critical role in advancing our understanding of SWS. Researchers have used mouse models to replicate the genetic mutations found in SWS patients, studying the impact on brain development and blood vessel formation. In one study, researchers used a mouse model to demonstrate that inhibiting the activity of the GNAQ R183Q gene could prevent abnormal blood vessel development in the brain and skin, suggesting a potential avenue for treatment.

Zebrafish models have also been used to study the development of blood vessels in SWS. Researchers have found that zebrafish share many genetic similarities with humans,

making them ideal models for studying genetic mutations. In one study, researchers used zebrafish models to identify a new drug candidate for SWS. The drug, called rapamycin, was found to reduce the size of abnormal blood vessels in the zebrafish model, offering hope for potential treatments in humans.

Animal models are not only critical for studying disease mechanisms and potential treatments, but they also play a crucial role in drug development. Before a new drug can be tested in humans, it must go through rigorous testing in animal models to ensure its safety and efficacy. Animal models can provide valuable insights into potential side effects, optimal dosing, and drug interactions, speeding up the drug development process and bringing new treatments to patients faster.

“Animal models, particularly mice and zebrafish, have played a critical role in advancing our understanding of SWS.”

In conclusion, animal models, particularly mice and zebrafish, are essential tools in rare disease research. The use of these models has advanced our understanding of rare diseases, including Sturge-Weber syndrome, and has led to new treatment options for patients. While animal models cannot fully replicate the complexity of human diseases, they provide a critical foundation for further research and drug development. The continued use of animal models in rare disease research offers hope for patients and their families, and the potential for new and effective treatments in the future.

What's Next and How Can You Help?

The late **Kimberly Slate** told us her story in 2018. In the closing paragraph she said,

“Life is uncertain. We are all dealt a deck of cards and some of us have more cards stacked against us than others, but we are all human. There are universal feelings we will all experience at one time or another. Please believe, that life is a journey with unforeseen forks and speed bumps but together as a unified community together we are here to cheer each other on; to provide support when others can't comprehend what we go through. Together we are strong. Together we can stand tall. Together we are one.”

Each year **The Sturge-Weber Foundation Month of Awareness** rolls around in May. It is a chance for us to UNITE with one another. To spread awareness of SWS, KT, and PWS like dandelions. It is the one month when we are uniquely and firmly together, standing strong.

Since before the discovery of the GNAQ R183Q gene mutation 10 years ago, The Sturge-Weber Foundation has been working hard to support patients, caregivers, researchers, and doctors on a united front.

The journey is not over. Not until there is a cure. The sad fact is research takes money and resources. With help from warriors like yourself we can achieve our mission. **With just \$10 a month (for the GNAQ R183Q 10 year anniversary) we would be able to accomplish funding for vital research beyond discovery of this gene.** We could find the “what's next” piece of the puzzle!

Read Kimberly's Garden Memorial on the next page.

GOAL: \$100,000



GNAQ \$10
RECURRING DONATION *Challenge*

What can \$10 buy? Two tall lattes at Starbucks®? A combo meal at your favorite fast food restaurant? Or, a \$10 recurring monthly donation, **in honor of the 10 year anniversary of GNAQ R183Q discovery**, to The Sturge-Weber Foundation will last longer than 15 minutes it takes for you to drink your morning cup of joe—and it is guaranteed to help more people!

It's been 10 years since GNAQ R183Q was discovered but there is still so much to do! Will you unite with us to find “what's next” in SWS research?

Click the QR code at the right or go to www.Sturge-Weber.org. Scan (or click) the **Donate** tab, click **Recurring Donation**, Click **Other Amount** and type in \$10. You can choose the date your donation is given each month. Complete the rest of the form and you are done.



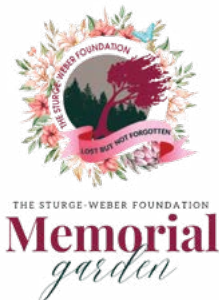
ARE YOU UP FOR THE CHALLENGE?

[Donate Today!](#)

Until We Meet Again... Remembering Kimberly

Kimberly burst into our lives and, in typical fashion, jumped in with both feet as an appointed member of the SWF Patient Engagement Program. Her luminous smile and hearty engaging laugh made everyone feel at home as they convened to set SWF engagement strategies for clinical care and research. Her inquisitive mind and willingness to help wherever needed were so cherished at a time when we were just building the program for adults living with SWS.

She loved life with a ferocity like no other draining every last drop out of it! She now has a much grander view over all of us which means watch out our world will never be the same and so much richer for her presence in our lives.



Visit the SWF Memorial Garden
online: Sturge-Weber syndrome.

www.sturge-weber.org/who-we-are/swf-memorial-garden

Or, contact Julia for more information,
jterrell@sturge-weber.org

Mark Your Calendars

Educational Mini Summit

12:00–3:00 pm EST
June 3 and August 5

[Learn more](#)

Million Miles Walk

Now–July 19, 2023

[Get Involved](#)

Town Hall Meeting with Karen

May 24, 9:00–10:00 EST

[Learn More](#)

SWF Month of Awareness

The entire month of May

The SWF Day of Giving: May 25

[Learn More](#)

Branching Out

In-Person Educational Day

Boston Children's Hospital, May 20
8:00 am–4:00 pm

[Learn More](#)

Running with the Sharks

June 11, Beach Haven, NJ

[Sign up here](#)

CCN/SWFIRN Meeting for Professionals

September 14–17, 2023

Renaissance Hotel in Charlotte, NC

[Sign up here](#)

Coming Soon

October we are going to CA, TBA

Town Hall Meeting with Karen

November TBA

The Importance of Tissue Donation

Barbara Jean Osborn's Story

Republished from *Roots in Research*, 2020



Barbara and Karen

I, Barbara Jean Osborn, was born on February 22, 1937. I was born and was raised up on a farm, life where the medical situation was nothing compared to what it is today! My reason for bringing this point to the table is due to the fact I was born with, what I now know as Sturge-Weber syndrome (SWS), back then, we knew it as a birthmark.

As I grew older and attended school I was not only teased by other students but I was told it was an angel that had kissed me on the face. My mother was always working so there really wasn't anything she could do to help as far as going to doctors. My father was too busy working then going out drinking afterwards.

My older siblings were always there to assist and help until I was old enough to take care of myself. I have had a couple of occasions where my younger sibling and I were sitting at our kitchen table when I was approximately 7-years-old and I was told I had a seizure and blacked out and fell onto the floor. There was no convulsing, just a blank stare and I woke up on the floor. Another time I was told that I was outside playing and became very tired so I went to sit down and I had a blank stare but didn't black out.

Skipping ahead several years to my adulthood, I have found myself in my elder years not becoming very good friends with SWS. In

my elder years I have experienced more dramatic symptoms such as: TIA'S (mini strokes), blindness in my right eye, congestive heart failure, kidney disease and several bad cases of pneumonia. I have fought this so far for 83 years. It has not been an easy road.

Thank you & God Bless!

Barbara Jean Osborn

“...I have found myself in my elder years not becoming very good friends with SWS.”

A Note from Julia:

Barbara and Karen have been fantastic to work with through their journey. Sadly, Barbara lost her battle and went home to heaven. Even in the end she wanted to further the science for Sturge-Weber syndrome with hopes that others would not have to suffer as she did.

Today let's honor Barbara and her family by signing up to donate tissue. Email me today and I can help you at jterrell@sturge-weber.org. Our hearts go out to the Osborn family. Barbara was a true Warrior.

READ ROOTS IN RESEARCH REPORT [Here](#)

THE SWF APPROVED TISSUE DONATION
COLLECTION CENTER:



Reunion of Champions honoring Mathew Avram, MD



NEW ORLEANS · 2023
REUNION OF CHAMPIONS
Laissez les Bon Temps Rouler

On March 17, 2023, The Sturge-Weber Foundation traveled to New Orleans, Louisiana to honor **Dr. Matthew Avram, JD** at our **Reunion of Champions** Event. The event was attended by dermatologists from all over the country. Our sponsors were very generous from **Sofwave, Candela, Blossom Innovations, Laser and Skin of New York, Lutronic, and Burkhart**

Pediatric and Adolescent Dermatology. The event was themed in St. Paddy's Day attire and decorations. The night would not be complete with a little fun too.

Dr. Avram was a great sport when roasted by **Dr. Brian Biesman** and **Dr. Jeremy Green** who were amazing. A special shout-out to our board members **Molly Speer, Crystal Elliers** in attendance and **Michelle Mora** and her husband **Jacob** who were all able to attend and support this wonderful cause.

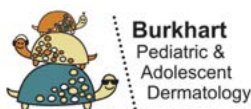


THANK YOU!

PLATINUM PARTNER



SILVER PARTNERS



MCs:

Dr. Brian Biesman
Dr. Jeremy Green

Location:

The Chicory

Revelers/Jugglers:

Frenchmen Street Productions

Balloon Art:

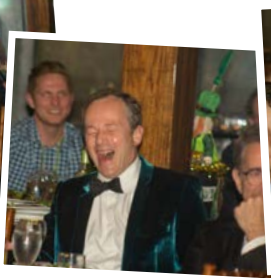
FunkShui Balloon Collective

Party Planner & Photography:

Rachelle and Ricardo at Leave It To Us Events and Celebrations

DJ/Music:

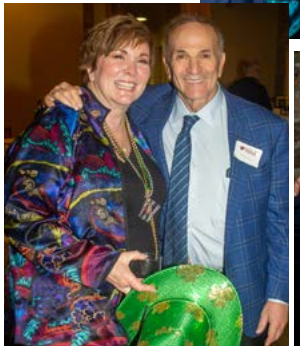
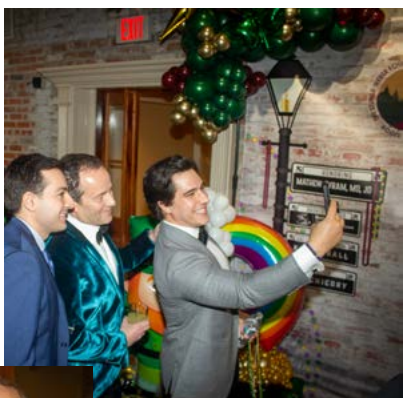
Calfee Productions



THE STURGE-WEBER FOUNDATION
 REUNION OF
CHAMPIONS
 NEW ORLEANS · 2023
Laissez les Bon Temps Rouler

Honoring:
 Mathew Avram, MD, JD
 (more photo on page 25)





STRONGER BECAUSE WE ARE



THE STURGE-WEBER FOUNDATION
BIRTHMARKS | GLAUCOMA | SEIZURES

Are you ready to lace up your walking shoes and pound the pavement for SWF? We are counting A Million Miles (collectively) for The SWF!

Does setting up a fundraiser sound intimidating? Well, we're here to tell you that it doesn't need to be complicated to be effective.

For 35 years the Foundation strives to find answers, improve medical treatments and forge opportunities in research to combat this rare disease that affects only 2% of the population.

IT IS THE FOUNDATION'S HOPE THROUGH THE MILLION MILES WALK EVENT TO:

- Raise funding for research
- Provide Adults with SWS support and retreats just for them
- Help fund our new online educational program for 2023

The SWF Million Miles Event starts now and ends July 19, 2023.

Create a Team, register as part of an existing team or join The SWF Team, go for a virtual walk, or walk as an individuals. It's all about the journey to a million miles and fellowship with one another.

You can choose to go big for The SWF like **Myla's Mission** and **Hailey's Hope for SWS** or you can keep it simple. Draw inspiration with the ideas on **The SWF Month of Awareness Action Calendar** on page 17.

2023 SHIRT DESIGN



Pledge It to raise \$100 and receive your SWF t-shirt!

New this year:

YOUTH, TODDLER, AND INFANT SIZES

REGISTER TODAY:

sturge-weber.org



OUR SPONSOR:



Inspired by patients.
Driven by science.

Spring 2023

Members Taking The Lead



Hailey's Hope

 Haileys-Hope-for-Sturge-Weber-Awareness
 haileyshopesws
www.haileyshopesws.com



Myla's Mission

 mylasmissionforSWS  mylasmissionSWS
www.mylasmissionsws.com

THINKING OUTSIDE THE BOX FUNDRAISING IDEAS



- Set up a virtual JustGiving page to collect donations.
- Ask local business owners if you can sit out a jar to collect spare change, decorate the jar for Sturge-Weber.
- Host a garage sale to support SWF.
- Ask friends and family to donate used books for a book sale with the proceeds going to SWF.
- Consider a Facebook Fundraiser.
- Work with a local business to host a car wash.
- Coordinate efforts with a local Karaoke for an “open mic night” to benefit SWF.
- Pancake Breakfasts are always yummy. What organization can you pair up with to host a fundraiser for SWF?
- Ask local businesses how you can host a fundraiser at their establishment. A local brewery, ice cream shop, coffee shop, or taco restaurant for a portion of proceeds on one day’s sales for SWF. Do your part to promote the event to make it a success.

Need more help?

Email us at swf@sturge-weber.org.
We are here for you!

Educational Mini Summit Recap

On April 1st we had a wonderful Mini Summit sponsored by UCB. We had 17 families attend. The topics of this mini summit are as follows:

Dr. Aimee Luat, MD from Detroit Children’s of Michigan discussed the history of neurology and what to expect. Dr. Elena Bitrian, MD from Bascom Palmer discussed glaucoma. **Dr. Sarat Thikkurissy, DDS** or Dr. Bobby told us all about dentistry and why it is important. He always says if you need recommendations to let us know. **Mitra**

Habibi, Pharma from UIC told us all about pharmacology and why it is important to understand what you take and how they interact with each other. Last but not least **Dr. Craig Burkhart, MD** from UNC and is now in private practice told us about lasers and skin creams as well as sunscreens. Great questions too ensued after each talk. These talks will soon be on YouTube. Don’t hesitate to check it out in June when we will talk about Assistive Technology, neuropsychology and much more.

OUR SPONSOR:  Inspired by patients.
Driven by science.

Continued from page 3
the Journey...

me on a conference call they'd found the SWS gene. Crystal clear. For once in my life, I was speechless! The journey is sweeter with our warriors beside us.

History can bring humility. History is our touchstone. It provides context—and inspiration. When we think back to leaving college or getting that first job, we remember how uncertain it felt. So much excitement and maybe anxiety. An SWS diagnosis brings dark days yearning for light and joy. The death of loved one's agony yet peace.

History gives us an anchor, so we remember our mistakes to never repeat and what to never do or say again. It's not about how big a load we carry or how long we can soldier on—but just the opposite. We see how truly fleeting a life can be and how fragile life can really be. Humility brings hope which leads to joy. We are meant to live in J O Y!

Ambiguity is OK. Daddy said, "Karen, you have enough of a load to carry with Kaelin". You gotta take it slow. It's too challenging"

But whenever a challenge crossed my path, I had their example of how to face adversity. Head on. Head held high. Nose to the grindstone when necessary. And ALWAYS with a wicked sense of humor! After all, it takes courage to challenge old ways of thinking and forge new paths for exploration and confirmation. A willingness to not be unsettled by ambiguity because you know the path is ahead and the outcome already ordained. **It's a journey.** The fact is you don't recognize sadness without knowing joy. You recognize the darkness because you have appreciated the light and love of those around you. And with hardship, we dust ourselves off or maybe pat ourselves on the back, but we can and will rise again. Quitting is NOT an option! Indeed, that's what—and why—we remember.

Seek joy and embrace it!

Karen L. Ball

Continued from page 4
GNAQ R183Q...

the syndrome, efforts will likely be directed towards developing more precise and effective therapies that target the mutated gene. Furthermore, the use of emerging technologies such as gene editing and improved targeting of pharmaceuticals may hold promise for developing individualized treatments tailored to the specific needs of each patient. Thanks to the dedication of researchers, clinicians, and patient advocates, we are making significant progress in understanding the disease biology of SWS and developing new tools to study and treat this condition. The Sturge-Weber Foundation and its partners, as always, remain committed to supporting ongoing research and advocacy efforts to improve the lives of those living with SWS.

READ THE ORIGINAL REPORT:
www.nejm.org/doi/full/10.1056/nejmoa1213507

The SWF Official YouTube Channel

SWF TALKS

The Sturge-Weber Foundation

WHY WE DO WHAT WE DO.
with Kaelin Ball

More information:
www.sturge-weber.org

Have you had a chance to watch our "SWF Talks" on YouTube yet? You can watch talks with Kaelin Ball, Dr. Dave Shahani, the Speer Family, and more!

Be sure to head over to, as they say, "like and Subscribe to our channel" today!

[@TheSwffoundation](https://www.youtube.com/@TheSwffoundation)

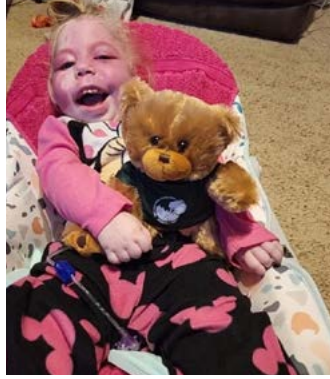
LIKE & SUBSCRIBE

Special Delivery by Webster Bear

One of our programs is to send our mascot, Webster all of the world for cuddles and comfort. We love to see the joy he brings to everyone's face! If you, your child, or someone you know who has Sturge-Weber syndrome and is getting any type of treatment and feel they could use the comfort of Webster bear, just let us know!



Calvin



Emersyn



Jax



Sadie



Kingsley



Danica

ON THE ROAD *with Karen*



Karen just attended the annual meeting of the American Society for Laser Medicine and Surgery INC (ALSMS) in Pheonix, AZ.

There she got to meet with the team working with **Dr. Kristen Kelly** from our Clinical Care Network, **UC Irvine** as lead investigator. The study they are working on is called Optical Coherence Tomography Skin Imaging and Measures and you can find more details at <https://clinicaltrials.gov/ct2/show/NCT00764920>.

Photo by Karen Images



BURGUNDY MAY

The Sturge-Weber Foundation Month of Awareness

www.Sturge-Weber.org

Preparing for May Month of Awareness

The SWF Month of Awareness starts May 1st. This is our opportunity to spread awareness and help fund important research, programs, and education.

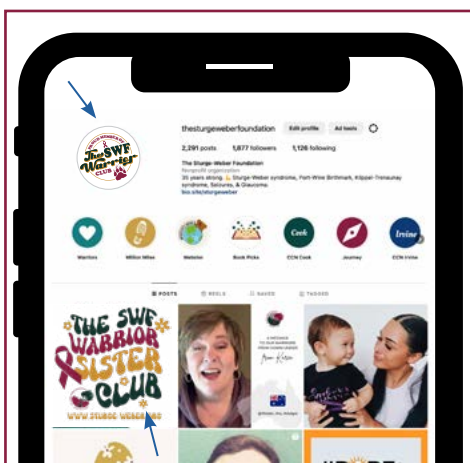
Even though it's the 21st century with so much advanced technology and vehicles for communication, Sturge-Weber syndrome is still unknown to so many. Education and awareness eliminates fear and stigmas.

Did you know that YOUR personal story is the best way to shed light on this disease?

This is YOUR month to share YOUR story in YOUR community, whether your community is on social media or around your neighborhood.

We can change the face of SWS together because we are united.

Click the link below to grab your free SWF Month of Awareness graphics and profile



[Get your FREE social media graphics for Month of Awareness here.](#)

photo. Post them on social media to show your support and add a story of your journey in the caption.

TikTokers Unite. We are also looking for anyone willing to tell their story through the power of TikTok. That's right, The SWF has joined TikTok to educate a whole group of viewers! We couldn't be more excited at this opportunity.

So much of our progress comes from all of us being a caring and sharing community. Across the globe we can be THE ADVOCATE for all those with SWS and be the source of improved quality in their lives! What an amazing time to be UNITED!

Don't be shy. If you are interested in filming short videos for us, let us know. Contact Julia at jterrell@sturge-weber.org.

Together, we are stronger because we are united.



[Visit the Official SWF Shop for more gear here.](#)

SAVE 10% ON YOUR ORDER!



MAY 2023

SWF MONTH OF AWARENESS ACTION CALENDAR

MON	TUE	WED	THU	FRI	SAT	SUN
<p>7</p> <p>Host a restaurant fundraiser and advertising on social media and flyers.</p>	<p>1</p> <p>Use one of our free social media graphics to post and show your support for SWF.</p>	<p>2</p> <p>Ask family and friends to pledge for your miles walked the month of May for the Million Miles Walk.</p>	<p>3</p> <p>Share your SWS journey with those online. Hint: always end with an ask for donations.</p>	<p>4</p> <p>Share a SWF post to your social media feed(s), don't forget to tag us.</p>	<p>5</p> <p>Ask local businesses to post our poster in their window or on a community bulletin board. <i>Download and print.</i></p>	<p>6</p> <p>Host a restaurant fundraiser and advertising on social media and flyers. <i>Krispy Kreme, Home Ica, Little Ceasars, to name a few.</i></p>
<p>14</p> <p>Organize a car wash with your group to benefit the SWF and raise awareness, don't forget the sunscreen!</p>	<p>8</p> <p>Host a summer kick off fun run or walk at your local park as part of the Million Miles Walk (MMW).</p>	<p>9</p> <p>Challenge your gym buddies to a treadmill challenge. Also counts as MMW.</p>	<p>10</p> <p>Host a bake sale in conjunction with a local car show.</p>	<p>11</p> <p>Cyclers, get your buddies together for a ride for SWF and collect pledges.</p>	<p>12</p> <p>Turn your walk into a dog walk, encourage the dogs to dress up, and collect pledges.</p>	<p>13</p> <p>Host a BBQ in your backyard, invite everyone you know, and charge a cover charge to benefit SWF.</p>
<p>21</p> <p>Ask local businesses to put a donation jar on their counter. Extra change can add up!</p>	<p>15</p> <p>Host a block party with games and food. Charge admission for donations.</p>	<p>16</p> <p>Create a facebook fundraiser, it's super easy and free.</p>	<p>17</p> <p>Company owners, become a corporate sponsor of the MMW.</p>	<p>18</p> <p>Make up your own.</p>	<p>19</p> <p>Get your community involved, ask organizations to collaborate for a fundraiser.</p>	<p>20</p> <p>Does your company match donations? Make sure to add that to your donation.</p>
<p>28</p> <p>Clean out your closet and organize a yard sale with proceeds benefiting the SWF. Ask friends to add items to sale.</p>	<p>22</p> <p>Book-aholics, sale your used books for donations and make room for more books.</p>	<p>23</p> <p>Create a Reel or TikTok to spread awareness of SWS. Have fun with it!</p>	<p>24</p> <p>Jeepers, create a Jeep ride for SWF. Create SWF flags, decorate your Jeep, gather donations.</p>	<p>25</p> <p>Write letters as part of your fundraising appeal. Make it personal. Tell them why they should donate to SWF.</p>	<p>26</p> <p>Create an outdoor movie night with family and friends. Charge a donation entrance fee.</p>	<p>27</p> <p>Parents, share one thing you wish people knew about SWS on social media.</p>



STRONGER BECAUSE WE ARE
UNXITED

www.Sturge-Weber.org

The Aussies are Coming!

Part of The Sturge-Weber Foundation's mission is to expand our worldwide membership and support international SWS organization's missions as well.

One of those organizations is "Weber The Wedgie, an organization that raises fund to help support Australian families living with Sturge-Weber syndrome.

Founder Michael Duffy says, "I managed to raise my target of \$30,000 which is amazing, so we work on getting as many Aussie families across to the conference [in 2024] as possible."

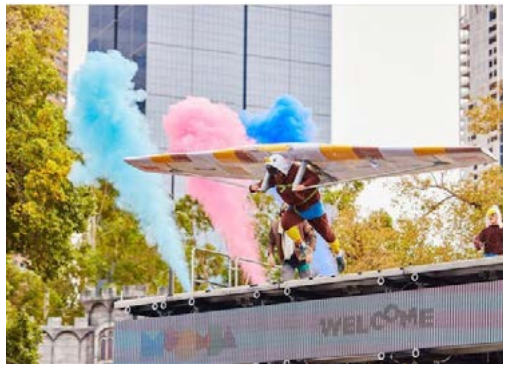
WOW! Michael, we cannot wait to see all the families from down under, mate!
In a recent event called the **2023 Moomba Birdman Rally**, Weber the Wedgie Team placed 2nd with an awarded cash prize of \$5,000. All they had to do is strap homemade gliders on their back and "fly" off the platform with the Yarra River awaiting below and compete to see who can "fly" the furthest – all in the name of charity.

The Moomba is Australia's largest free community festival held during Labour Day Weekend in March. Which sound like a blast!



Connect on Instagram

 [@weber_the_wedgie](https://www.instagram.com/weber_the_wedgie)



Resources

Facebook Groups

OFFICIAL STURGE-WEBER FOUNDATION AWARENESS PAGE:

A public place created to share your questions, stories, pictures, and videos.

[sturgeweberfoundation](https://www.facebook.com/sturgeweberfoundation)

SWF ROAD WARRIOR

BEAR WEBSTER:

Webster is our Road

Warrior who loves to get his picture taken.

[SWFRoadWarriorBearWEBSTER](https://www.facebook.com/SWFRoadWarriorBearWEBSTER)



STURGE-WEBER FOUNDATION WORLD OF CARE AND SHARE NETWORK:

A place to gather so we as a group have an open forum to ask questions, learn together and brain storm.

Let's Connect



Sturge-Weber Syndrome Family Education Day

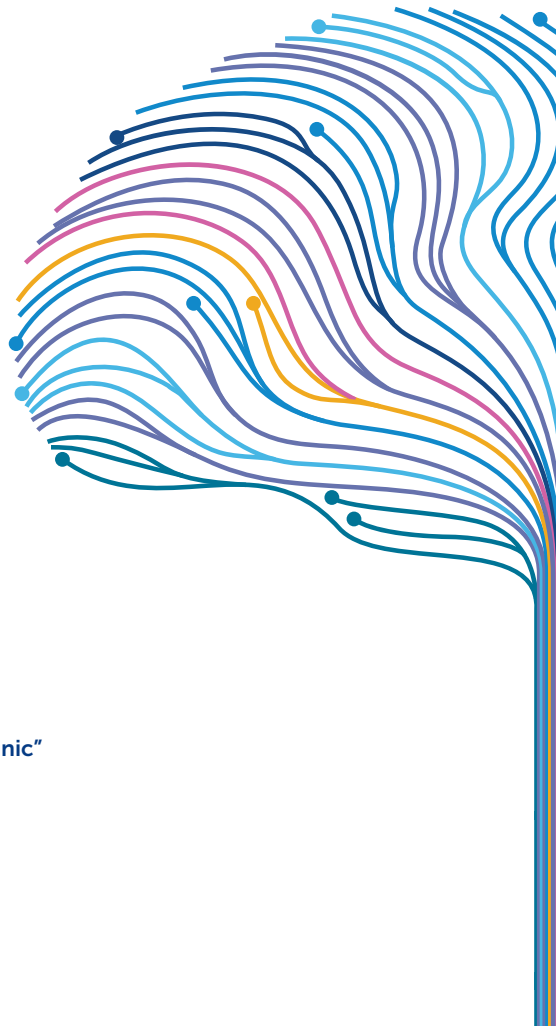
hosted by Boston Children's Department of Neurology

Saturday, May 20, 2023

Longwood Hall - 342 Longwood Avenue, Boston MA 02115

Schedule of events

8–9 a.m.	Registration
9 a.m.–noon	Lectures
9–9:20 a.m.	Welcome and TNC Introduction Mustafa Sahin
9:20–9:40 a.m.	Gene and Mechanism Matt Shirley
9:40–10 a.m.	Ophthalmology Ankoor Shah
10–10:20 a.m.	Epilepsy Surgery Approach Michelle Chiu
10:20–10:40 a.m.	Neuropsychology Katrina Boyer
10:40–11 a.m.	Coffee Break
11–11:20 a.m.	Neuroimaging Csaba Juhasz
11:20–11:40 a.m.	Vascular Biology Research Joyce Bischoff
11:40 a.m.–noon	Current Trials/Treatment Anna Pinto
Noon–1 p.m.	Lunch
1–3 p.m.	Meet the Experts–“Super Clinic”
3–4 p.m.	Vascular Biology Lab Tour
4–5 p.m.	End of Day Reception



Boston Children's Hospital
Neurology

[REGISTER HERE](#)

Julia Selected to be Part of the Rare Compassion Program



JULIA TERRELL
The SWF Director of Community Relations



ERIK SOUTHARD
First medical student from University of Toledo

We are excited to announce our own Julia Terrell, Director of Community Relations was selected to be a part of the **Rare Compassion Program from Global Genes**. The RARE Compassion Program provides an opportunity for medical students to learn about the unique needs and challenges individuals and their families face living with an undiagnosed or rare disease (such as SWS). Julia was paired with, **Erik Southard**, a first year medical student from University of Toledo. The first conversation was very enlightening and was exciting to explain our syndrome and Erik explained how he was interested in neurology and psychology. The program will last four months and we will be in constant communication over different topics. Stay tuned for more information to come.

Month of Awareness Instagram Giveaway



The Sturge-Weber Foundation is excited to giveaway a copy of “*Beautifully Blemished: Learning and Celebrating Skin Differences*” a children’s picture book by Leanne

Stuckey, AND a gift box for The SWF Month of Awareness!

You don’t want to miss it! **Follow us and Beautifully Blemished on Instagram** now so you are ready! One giveaway at the beginning of May, and a second around The SWF Giving Day on May 25th.

About the book: Having grown up with a visible skin difference, I understand the

feelings of insecurity that come along with that reality. This book was written to help kids truly recognize the beauty in their uniqueness— hopefully much sooner than I did. The goal is to empower young boys & girls, while spreading awareness and helping to shift society’s narrative of what beauty is and what it looks like. Boxes by Loved Well Boxes.



Follow us today!

 [@thesturgeweberfoundation](https://www.instagram.com/thesturgeweberfoundation)

 [@BeautifullyBlemished365](https://www.instagram.com/BeautifullyBlemished365)

Disclaimer: Giveaway will be announced on Instagram. You will need to follow the instructions on the post. Giveaway will not be available on Facebook. Winners will be drawn at random using a giveaway app.



COMING SOON: Warrior Mama Seal of Approval



Dr. Craig Burkhardt and Tynan

How many physician's waiting rooms have you sat in related to your SWS diagnosis? How many times have you had the most compassionate support and stellar communication and service? I suppose we have all had educators, healthcare providers and more who simply did their job or felt compelled to minimize what you've learned along your SWS and birthmark related journey. The patronization or lack of communication one receives is always easy to tell others, "Oh, do NOT go to xyz provider because..."

It is always the goal of the SWF to enlighten and advocate for each of you always. We aim to focus on the positives and those who uplift us in this journey which is already tough enough without being left feeling demoralized or unheard by a healthcare provider. We ask that you continue to let us know of referrals that maybe weren't a right fit for you or one that you had some complaints about!

Which teachers will be remembered for life? We like to highlight and praise everyone who uplifts us on the journey; those who had excellent communication and administrative staff. Thanks to a GREAT conversation with a Warrior Mama named Lina we have decided to launch the **Warrior Mama Seal of Approval!** The seal is awarded to those healthcare providers and teachers who go above and beyond to give compassionate and excellent service and who have your six (aka "who have your back") as they say in the military! I'm proud to announce the first wave of seals have been awarded in Dermatology.

The Reunion of Champions honorees and their staff are examples of who we want on the front-lines of this war we're waging

against SWS and birthmarks.

This is criteria in which we base our Warrior Mama Seal of Approval:

Administration

- Ease of appointment scheduling
- Flexibility on changes
- Staff demeanor and attitude
- Quality of the billing process
- Charges for service

Communication

- Effective and relateable communication
- Willingness to dialogue until all questions answered or set up follow up time to address them
- Acknowledges parental/guardians understanding of SWS and related matters as a warrior on the front-lines living with it
- Process to address miscommunication
- Willingness to accept responsibility for actions

Expertise

- Certifications
- CMEs
- Interest in gaining more knowledge on SWS related matters
- Conduct more impactful research

Access to Care

- Willing to write letters of medical necessity for insurance issues
- Referrals easy to obtain
- Works within and without their institution to work in tandem with you on any issues

We look forward in the coming months to tell you about this amazing program to recognize outstanding providers of healthcare and support to our SWS patients. We are on a journey of discovery and support for all!



#SWFNOLA2023 REUNION CHAMPIONS THE SWF-NOLA

#SWFNOLA2023 REUNION CHAMPIONS THE SWF-NOLA

#SWFNOLA2023 REUNION CHAMPIONS THE SWF-NOLA '23



STRONGER BECAUSE WE ARE



Your donation is driving change

- YES!** I will make a tax deductible donation of \$_____.
- YES!** I want to make an ongoing monthly pledge of \$_____.
- In **MEMORY** of: _____
- In **HONOR** of: _____
- Endowment Donation
- Stock Donation
- I have enclosed a check
- Please pay by credit card* VISA MasterCard Amex Discover

Name on Card: _____

Credit Card #: _____

CSV: _____ Exp. Date: _____

**You may also make a secure donation at www.sturge-weber.com/donate or scan the QR Code below.
Please provide your contact information in the area above for credit card donations.*

***You may also **donate gifts of stock or real estate** to the Sturge-Weber Foundation.*

Name: _____

Address: _____

City, State, Zip: _____

Email: _____

Mobile Phone #: _____

By providing your email and mobile number, we are able to send you news updates immediately!

SAVE THE STAMP!

**Use the QR Code to the right to donate
securely on-line
or fill out form and mail to:**

The Sturge-Weber Foundation
6105 S. Main Street, #200
Aurora, CO 80016



sturge-weber.org/donate-today