

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



Branching Out

FALL 2023

LEGACY LEADERS

planting seeds for a fruitful tomorrow



What is your Legacy?

leg-a-cy /le-ges-ē/ n. the long-lasting impact of particular events, actions, etc. that took place in the past, or of a person's life.

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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

FALL 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

Karen's 2 Cents

Preparing for the Storms of Life

Today started out like any other day when I'm with my Daddy. I made breakfast and cleaned up the dishes as we got ready to go into Kalispell, MT (population 24,600). He drove because he was older and wiser and had a great sense of direction. I had some shopping to do while he ate lunch with his buddies, so I took note of the sign on the restaurant so I'd know where to go back to get him.

I've navigated the roads in Kalispell for over 30 years since my parents retired to Bigfork, MT, so I didn't think about driving off to shop!

I head off to the local Walmart to buy a few things and kill time. Then, I headed back to the restaurant and spent an hour driving up and down the streets looking for the restaurant!

I became increasingly frantic as I went up and down the blocks; I said a prayer and asked for a sign. I stopped at the police station and explained I couldn't remember the restaurant I left Daddy at, and the woman behind the counter, also named Karen, started naming off restaurants where he could be. Then, another woman who was there started googling and helping. It's incredible how small towns always come together to help each other. We (The SWF) lose that sometimes in a big city.

"You never know when a crisis or storm will come and how ferocious or unexpected it will be!"

I calmed down gradually with their help. Committed to finding the restaurant, they suggested that I go around the backside, and lo and behold, there is my 93-year-old Army reservist, national guardsman daddy, sitting on the front stoop, waiting for his ride from his one and only daughter! (You may have guessed he had no phone and hadn't memorized my number.)

We both shuffle our way to the car, as tears slipping out of my eyes and him exhausted beyond measure. We made it home for him to "rest his eyes," and I said prayers! I was raised to always be honest and come clean. I can admit I have not always done that, but I always knew in the back of my head that honesty is the best policy.

Later, with a couple sips into our toddy on the deck, I ask him, "Hey, what would you do if I didn't come around for hours?" (Beverly Jean, his late wife of 70 years, passed away in April was always in charge of the cell phone and the direction in life. Neither of us gave much thought to the idea that maybe he should have a cell phone with him in an emergency.) Then I said, "Hey, I have a story. You'll never guess what happened today!"

I then regaled my story to him and came clean about the events of the day, and then, in

Continued on page 8 >>>

TEXT SWFUNITED TO 53-555 TO LEAVE A LEGACY OF HOPE.



A Great Year and So Much More Ahead

JEFF LOEB, MD, PHD
The SWF's Chief Scientific Strategist

Every year, I am asked to reflect on what we have accomplished and what lies ahead. It has been a great year, and so much lies ahead! Our first combined scientific and clinical meeting last September in North Carolina was a highlight of the year. Not only was it a great meeting, but we received a prestigious grant from the National Institutes of Health to support this unique meeting, which helped us bring together our brightest scientists and compassionate healthcare providers under one roof. We asked our scientists to understand the personal challenges of this disease, and we asked our clinicians to think about the underlying science behind the disease. Individuals and families with Sturge-Weber were there to tell their stories and generate momentum for all of us to push for a better understanding of the disease and new treatments.

We gave an update on our tremendous progress with our BVMC (Brain Vascular Malformation Consortium) project, also funded by the National Institutes of Health. I get the same question asked to me from concerned parents of babies who have just been diagnosed, teenagers who are maturing into adults, and adults who are getting older: 'What happens next?'

The BVMC project will help answer this question by enrolling people with Sturge-Weber into our longitudinal 'big data'

study. This study is still actively recruiting, collecting clinical records and MRI studies, and looking for blood biomarkers to help us understand what happens next, what treatments are working and which are not, and to be ready to test new treatments soon. As part of this project, we have built a really cool 'dashboard' that creates a timeline for

"We will need considerable funding and expertise to transform our research database into a useful patient-centered dashboard and sustain it by transferring it to the Foundation."

each person with Sturge-Weber. A significant goal ahead is to transition this from a big data/AI research platform into a clinically useful tool for improved care. We have recruited almost 100 patients but have a lofty goal to enroll many more. We are also looking for ways to continue and expand

this database and link it to our cell phones and doctors.

Everyone who participates will be helping many others now and in the future. So don't hesitate to contact our research coordinator, Luz Rosales (luzr@uic.edu), if you would like to learn more and hopefully join us!

Finally, we continue to work with all of our Clinical Care Network sites and our fantastic physician leaders in neurology, dermatology, and ophthalmology to translate our growing dataset from the BVMC project into a better understanding of new treatments. We continue to have successful mini-summits covering all sorts of topics, and we are primed and ready to start clinical trials to

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Research is Like a Detective, Uncovering New Possibilities in Rare Disease Treatments

We're pleased to share some essential discoveries from our research into Sturge-Weber syndrome (SWS), thanks partly to early The SWF grant funds and the highly related disease, Phakomatosis Pigmentovascularis with Dermal Melanocytosis (PPV-DM). PPV-DM, for those who don't know, is the same as SWS but has pigmentary problems, which can affect the skin, eyes, and/or brain. We and other dedicated research teams had previously spotted that these diseases are caused by 'mosaic mutations' - genetic changes that appear in some cells in the body but not all - in two genes, GNAQ or GNA11. To identify new potential treatments, particularly for children who experience early-onset neurological symptoms such as seizures and developmental delay, which worsen over time, we delved deeper into an intriguing clue in brain images known as "tramlining."

This distinctive calcium deposition pattern, seen in parallel lines within blood vessels, led us to hypothesize that calcium levels might be an essential part of the diseases. We examined 42 children thoroughly by analyzing the calcium profiles in their blood. Our results revealed that approximately 41% had slightly reduced calcium levels, and intriguingly, lower calcium levels (even within the normal range) were linked to seizures and some specific anti-epileptic medications, even in the presence of regular vitamin D levels.

We don't know if this association is because one thing is causing another; at the moment,

this is simply an observation. As we carried out repeated measurements, we uncovered a notable trend of fluctuating calcium levels in patients, so different measurements on different days produced different results. Still, across the cohort, a substantial proportion was always abnormal. The calcium level was not so low as to be concerning for health, and bone density scans in patients with low calcium levels were normal. However, when we closely examined brain tissue

from patients who had undergone epilepsy surgery, we identified calcium deposits within blood vessels, around small vessels, and inside brain cells. This neurovascular calcification was progressive over time when brain scans were reviewed. This has expanded our understanding of the

disease as it challenges the earlier belief that calcium deposits were simply a non-specific sign of brain damage.

Instead, they might play a central role in the neurological progression of this condition. In parallel, we tested what was happening in the cells with these gene mutations in the lab. We discovered a continuous release of excessive calcium within these cells, much like an "always on" calcium switch. This overacting calcium signalling was so strong that it led the cells to pull in even more calcium from outside through transporters on the cell membrane called CRAC channels. We do not yet know if this calcium signalling problem is directly related to the findings in the blood of the patients or not. It is possible that. We aimed to find a way to "turn off" this overactive signalling; to achieve this; we

"This has expanded our understanding of the disease as it challenges the earlier belief that calcium deposits were simply a non-specific sign of brain damage."

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Making Strides in Research

MATT SHIRLEY, PHD
SWF Chief Science Officer

As we reflect on the strides we made in 2023, it's with great enthusiasm that I share the remarkable achievements of the **Sturge-Weber Foundation International Research Network (SWFIRN)**, particularly focusing on the groundbreaking discoveries unveiled at our combined clinical and scientific meeting this September in Charlotte, North Carolina. Our commitment to fostering collaboration between scientists and healthcare providers reached new heights, epitomized by this group of scientists and compassionate healthcare professionals, alongside individuals and families directly affected by Sturge-Weber Syndrome. The synergy generated in this unique setting fueled many exciting discussions and laid the groundwork for transformative therapeutic advancements.

Some of this meeting's many highlights came through presentations from junior scientists, showcasing novel insights into the vascular biology of SWS. Their work not only expands our understanding but also serves as a testament to the dedication of the next generation of researchers in our field. **Researchers from Japan's Nara Medical**

University presented work on microfluidic devices, creating a "vasculature on a chip" to study GNAQ mutant blood vessels in SWS. This innovative approach opens new avenues for studying the intricacies of the condition and devising targeted interventions.

Advancements in zebrafish models of Sturge-Weber Syndrome underscored the commitment to diverse research methodologies. These models offer valuable insights into the underlying mechanisms of the syndrome, paving the way for potential therapeutic breakthroughs. Collaborations with researchers from the uveal melanoma field brought a fresh perspective to our endeavors. Exploring shared interests in cell signaling and therapeutics, this partnership broadens the scope of our research, capitalizing on synergies between these related fields. The discussion of a recently published **mouse model** of Sturge-Weber Syndrome provided an opportunity to discuss a critical tool for further investigations and a platform for testing novel therapeutic interventions.

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Announcing Our New Clinical Care Network Member



Cleveland Clinic

9500 Euclid Ave S51 • Cleveland, OH 44195 • 216-445-7728
CCN Director: Dr. Ajay Gupta

Meet Reunion of Champions Honoree: Fernanda H. Sakamoto, MD, PhD



Dr. Fernanda H. Sakamoto, MD, Ph.D is an Assistant Professor in Dermatology at the Harvard Medical School, Wellman Center for Photomedicine, Massachusetts General Hospital, Boston, MA since 2006. She is a

fellow of the AAD, fellow of the American Society of Lasers in Medicine and Surgery (ASLMS), a member of the Société Française de Dermatologie; of the Brazilian Society of Dermatology, of the Brazilian Society of Dermatological Surgery, of the Brazilian Group of Melanoma and of the International Society of Dermoscopy.

She is a physician-scientist and board-certified Dermatologist from Brazil. Dr. Sakamoto's research interests include laser and device therapies, PDT, acne, skin cancer, wound healing, and new material science. Working with translational and clinical sciences in academia and industry is Dr. Sakamoto's field of interest. Currently, her work is focused on the development of new light-based treatments for acne and atopic dermatitis. Dr. Sakamoto has been a Co-Director of the Education Committee of the Wellman Center/MGH since 2017 and has served on the ASLMS Education, International Committees, Research and Development Committee, and Women in Energy Based Devices Committee of which she was the Chair in

2021. She was the Program Chair of the 2022 ASLMS Annual Meeting and has received the Best Overall Scientific Award, Best Clinical Science Award (twice) Best Basic Science Award, the Horace Furimoto Award for Innovation, and the Ellet Drake Memorial Award for innovative laser procedures by the same society.

Dr. Sakamoto has contributed with over 30 scientific and clinical publications in peer-reviewed journals, and over a dozen book chapters in the major Dermatology textbooks. She is an inventor on seven US and international patents on innovations in Dermatology. She has contributed to the creation of FDA-approved technologies, including the micro-coring device for skin tightening; and Dr. Sakamoto is the primary inventor of laser for acne treatment.

She has served as Associate Editor for the Journal of Investigative Dermatology for 4 years and was Guest Editor of Lasers in Surgery and Medicine in 2012. Dr. Sakamoto maintains a cosmetic laser/device clinic in Brazil, and as a founding member of the Dream Beam Foundation, she is actively creating a free clinic and training center in Sao Paulo. In the past, has treated native Indians in the Amazon jungle. Dr. Sakamoto also works as a consultant to multiple laser and cosmetic companies in the US. Dr. Sakamoto lives in Boston with her husband, Rox Anderson, her 3 boys under the age of 8 years old, and her little dog, BB. 🐾



REUNION OF CHAMPIONS

HALL
of Fame

FUNDRAISER KICK-OFF EVENT

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**"EVERY STRIKE BRINGS ME
CLOSER TO THE NEXT
HOME RUN."**

-BABE RUTH

HONORING

FERNANDA H. SAKAMOTO, M.D., PH.D.

MVP LEGACY LEADER: GARY LASK, M.D.

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Storms of life...

his inimitable way, he said, “Kar, if I called the policeman, he would say, what color is your car? I would say gray.” He said, “The Police would ask what kind of car you have – a Jeep. The Police would ask me your license plate, and I would say I don’t know!”

We both had a teachable life moment and a nice chuckle. Then he started talking about the storm coming in, making the pine trees sway to and fro. He said, “Kar life is like these storms. You never know when a crisis or storm will come and how ferocious or unexpected it will be!”

When people come together, we’re always uplifted and prepared because, just like these pine trees, you have some lower to the ground and those more mature and taller. We all come together in a storm; like today, the big ones protect the little ones beneath them and eventually pick them up.

That reminded me of our lives with Sturge-Weber, both for you and me. I like the unexpected storms that nature throws at us

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...A Great Year

optimize our treatments and explore new ones. I am particularly interested in treating brain calcification toxicity in Sturge-Weber. We are very close to launching a clinical trial to chelate this toxic calcium or prevent it from forming in the first place.

While we have successfully raised some funds to get this far, more is needed. We will need considerable funding and expertise to transform our research database into a useful patient-centered dashboard and sustain it by transferring it to the Foundation. Having this dream come true will improve clinical care and continuously feed new data into our database for future discoveries. We also need to fund new clinical trials that will

in Montana. It takes all of us in the forest to ensure the safety, security, love, and comfort of those just entering the forest and planting their seeds.

As we mature into fully grown evergreens, we spread our branches and guide and protect those, and as we become weaker and weaker through life’s challenges, those below us grow up to be strong and mighty Pines, oaks, and solid members of the forest.

P.S. In an emergency stop at the police station, saying help me. I’ve lost my Daddy, and I don’t know where to find him!

Legacy and “war stories,” as Daddy says, are built one moment at a time! Please join me in sharing your stories of overcoming challenges, giving up one cup of coffee or a hamburger a month, and donating to The SWF; just \$10 or more per month will build a legacy of hope and navigate us with knowledge. 🍀

With love, hope, and faith,



Karen Ball, CEO and Founder

dramatically change the course of the disease and make a life-long difference in those with Sturge-Weber.

So stay tuned, participate in our studies (PLEASE), and help us improve the lives of those who mean so much to all of us in another great year ahead. ✦

[LEARN MORE & REGISTER TODAY:](#)



Where are They Now?

Dr. Davide Zecchin of University College London the recipient of the 2018-2019 Lisa's Sturge-Weber Research Fellowship Award



During my PhD and first post-doc I was trained as a cancer biologist, and I studied the functional consequences of mutations occurring in cancers and approaches to treat

specifically mutant cells. Thanks also to a generous contribution from Sturge-Weber Foundation, in 2019 I joined the laboratory of Prof. Veronica Kinsler, where I had the opportunity to apply my experience to the study of the mutations that initiate Sturge-Weber syndrome. During our studies we realised that these alterations result in the dysregulation of specific signals inside the mutant cells. Our research, also propelled by a grant from the charity Sturge-Weber Italia, enabled us to uncover novel strategies for inhibition of these altered cellular signals. The next challenge

will be to translate these novel experimental approaches into treatments, especially to alleviate the neurological progressive symptoms that these patients often experience.

At that point, I came to realise that the very same genes that are mutated in Sturge-Weber syndrome are also responsible for initiating the most common ocular cancer in adults, called uveal melanoma. With a project aimed at applying the lessons learned from Sturge-Weber to uveal melanoma, I secured the funding support from Moorfields Eye Charity to start my lab at University College London-Institute of Ophthalmology. Back to cancer once more, but with a wider vision. I have recently embarked on this new challenge, and I am looking forward to building my research group further and providing my original contribution to advancements in this field. 🍎

THE SWF INTERNATIONAL FAMILY CONFERENCE



IT'S A July 11-14, 2024
WARRIOR
THING
PHILADELPHIA

"Our first conference was in 2017. It was really great to see the people you see on the Internet face-to-face and meet the families. I know in the first year or two of being diagnosed, going to in-person events are best."



Deborah Brewbaker



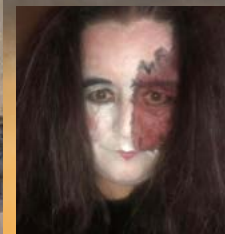
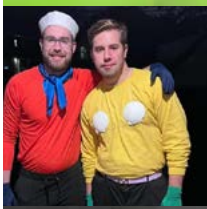
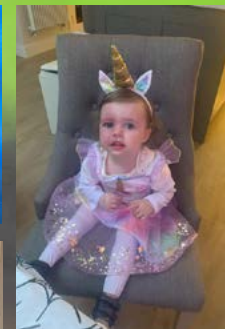
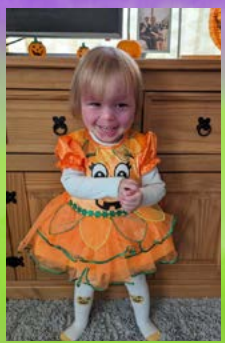
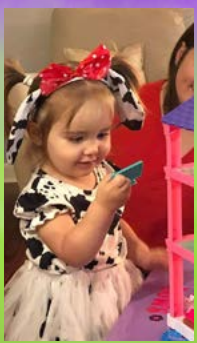
Wyndham Philadelphia Historic District

REGISTRATION OPENS SOON!

BACK TO SCHOOL



HAPPY HALLOWEEN



Partner Organization Spotlight: Patient Worthy



“Patient Worthy® is an online publication that provides relevant information to rare disease patients, caregivers and advocates alike. We connect those impacted by rare conditions with educational resources, advocacy groups, and news updates regarding rare disease research from across the globe. Our goal is to amplify the voices of those in the rare disease community, feature the institutions that serve them, and foster solidarity across diagnoses.

Here at Patient Worthy, if we are not learning to live with a chronic illness or rare disease ourselves, we are loving or working with someone who is. It is through this perspective, research and our passion for rare disease awareness that we bring you Rare Patient News. Well Done.” 🍀

Learn More: www.patientworthy.com

Languages supported on-site: English, Spanish, French, and Russian.

Podcast: WAIT, HOW DO YOU SPELL THAT? is a rare disease podcast produced by Patient Worthy.

Read or Submit a Story to Patient Worthy.



Animal Modeling

On Wednesday, November 8th, Davide Zecchin and Nicole Knoepfel presented their work with Veronica Kinsler to our very excited group. We had over 25 researchers present for an exciting conversation to finish out our year of research. More to come in the upcoming year.

I am thankful for...

“After using many eye drops for 14 years, my son does not need to use them. One year without eye drops.”

@rebecca_penders_mcdonald

I am thankful for...

“That my daughter is home recovering from Epilepsy Surgery”

@rebecca_penders_mcdonald



THE STURGE-WEBER FOUNDATION
6105 S. Main Street, #200
Aurora, CO 80016

Let's Connect



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Crick Research

explored different therapeutic approaches in the lab.

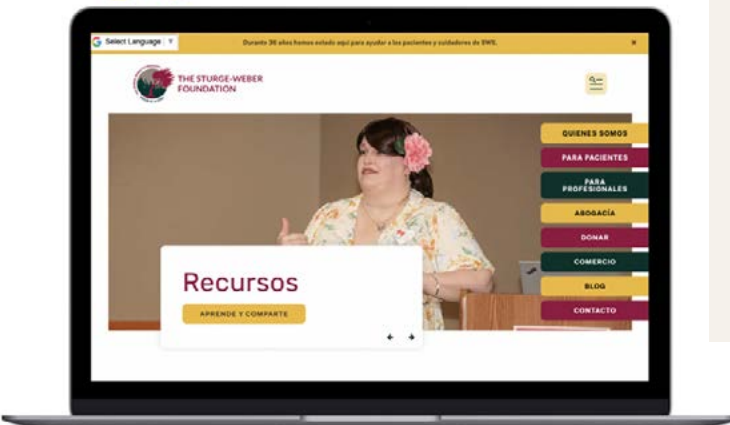
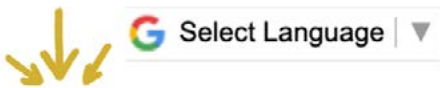
First, we developed specialized molecules, known as siRNAs, designed to silence these genes' "overactive" regions. Second, we used a drug that blocks the entry of calcium into cells through the specific CRAC channels. Both treatments improved the excessive calcium signalling calcium problems, but genetic therapy was the most effective. In conclusion, our research has pinpointed overactive calcium signalling as the primary problem in Sturge-Weber syndrome/PPV-DM. This breakthrough paves the way for new potential future treatments, either through genetic approaches or targeted

calcium drugs. These drugs are not yet available for these conditions, but we will continue working on this. 🍀

A research team at the Francis Crick Institute and Great Ormond Street Hospital (GOSH)/UCL Great Ormond Street Institute of Child Health have identified new potential treatments for children with rare genetic conditions of blood vessels, which cause severe, lifelong, and disabling symptoms like seizures and impaired development. Below are the findings. Read the entire research paper [here](#).

WE ARE MAKING OUR WEBSITE BETTER FOR YOU!

TRANSLATE INTO ANY LANGUAGE WIDGET



As part of our inclusive initiative, we are working to add a Google Translate button to our website. Stay tuned as we work hard behind the scenes on our accessibility improvements.

PEDRA in Atlanta



Pediatric Dermatology Research Alliance was hosted by our very own Dr. Esteban Fernandez, Dermatology Task Force Leader and Dr. Lisa Arkin, our CCN leader from University of Wisconsin. They both did an amazing job. We heard great talks on The Methodology of Innovation which included Dr. Rox Anderson, Dr. Blakeley, Dr. Garibyan, Dr. Gottesman, Dr. Lee and Dr. Levin. The keynote speaker Dr. John O'Shea from the National Institute of Arthritis and Musculoskeletal and Skin Diseases. I was also able to be a part of the Patient Advisory Committee which is so exciting to talk with other groups and work on topics like quality of life and more. There talks about birthmarks and patients' role in research. There might have been some

karaoke as well while we networked.

While in Atlanta I got to see many of our doctors from Clinical Care Networks around the country. It was so great to see Dr. Mo Levy, TX; Dr. Denise Metry and Dr. Reagan Hunt TX; Dr. Lawrence Eichenfeld, CA; Dr. Lisa Arkin, Dr. Beth Drolet and Dr. Todd Le, Wisconsin; Dr. Esteban Fernandez, OH; Dr. Anna Bruckner, CO; Dr. Megha Tolferson, Mayo Clinic; Dr. Deepti Gupta, WA; Dr. Cheryl Bryant and Dr. Mariam Igneibi, Cincinnati, OH; Dr. Ilona Frieden, CA. I was also able to get new contacts too. Can't wait to see what the future will bring and it was truly exciting to see so many wanting to know more about birthmarks. ●

Advocacy

Members of Congress are directly responsible for Federal programs that advance medical research, facilitate treatment development, and improve the quality and accessibility of healthcare.

The best way to ensure that Congress makes decisions, laws, and policies that empower our community is to use your voice and your story to personally educate your elected officials about the condition, the community, and contemporary issues. Connecting with your Members of Congress is easier than you might think. Below are the latest advocacy sign on we have been a part of:

NIAMS Coalition Clinical Trial Diversity Act Sign On: The Sturge-Weber Foundation, along with 43 members of the NIAMS (National Institute of Arthritis and Musculo-

skeletal and Skin Diseases) Coalition, representing millions of Americans with chronic conditions and diseases of the bones, joints, muscles and skin, as well as the professionals who serve them, undersigned in support of the National Institutes of Health (NIH) Clinical Trial Diversity Act (H.R. 3503).

Safe Step Act in PBM Reform, November 8, 2023: Over 100 patient and provider organizations, including The Sturge-Weber Foundation, are asking Congress to pass PBM reform and include the #SafeStepAct to help patients directly. The Safe Step Act (S.652/HR2630) could potentially be included in a PBM reform bill that is being worked on in Congress. ●

[READ MORE ADVOCACY HERE](#)

Hoedown to Throwdown for SWF

Howdy partners!! The Willard Family hosted a **Hoedown to Throwdown Sturge-Weber on Friday, October 6** at the Old Rugged Barn in **Towanda, IL**. Our event featured entertainment by The American Made Band, The Urban DJ, and line- dancing lessons. The cowboys and cowgirls were kept full and energized with pulled pork, mac-n-cheese and coleslaw provided by Woody and Bubba's BBQ. A popcorn bar and frosted sugar cookies helped satisfy sugar cravings and were both big hits. A Blackstone Griddle and a Solo Stove were our two big raffle items.



Attendees could also be "thrown" in jail and then bailed out for a donation and a Clear the Board (donation board) were additional ways we tried to raise more money at the event. Our own Sydney Willard and special guest, Karen Ball gave heartfelt speeches to the group and helped give everyone a feel for how their donations can help those living a Sturge-Weber life. Due to Covid, we had not hosted a fundraising event since 2019. Our 80's Retro Dance Party had raised \$5,170 and our goal with this fundraiser was to raise more money than the last one.



The Hoedown raised \$7,800. Our goal is to make these events bigger and better each time we have one which we all know is not an easy thing to do.



We cannot thank our family and friends enough for all of their help and support in making this a big success. Everyone who helped with planning, set-up, clean-up, donated food, drinks, made monetary donations and for everyone who attended, we are so grateful for each and every one of you. 🍎



YOU'RE INVITED TO OUR
TOWN HALL MEETING
with Karen
WEDNESDAY, DECEMBER 6 • 7:00-8:30 PM EST [Sign Up](#)

LEGACY LEADERS

planting seeds for a fruitful tomorrow

WHAT LEGACY ARE YOU LEAVING?

FOR Evan...Nicolly...the O'Reillys...



...all SWS members, researchers,
caregivers, and doctors.



**JOIN US AS A
LEGACY LEADER
TODAY!**

SCAN



DONATE **ONLINE** OR
TEXT **SWFUNITED** TO **53-555**

Planting a Legacy one acorn at a time

Why an acorn you may ask? Well, when Karen and Kirk Ball founded The SWF in 1987 they had a strong faith and belief that from little acorns grow mighty oaks! Technically, it was Stan Fisher who said it but he likes to share!

We have had so many of you join in your frightened early days of diagnosis and meet your new best friends, gain knowledge, find healthcare providers, participate in research and give back in the ways you were comfortable with in your corner of the world. To honor those who have built the foundation and grown our 'lil forest of mighty oaks, we are launching the **Legacy Leaders campaign**. By giving up a slice of pizza or a latte each month you can join us to build financial resources which keep the research momentum going, build educational resources through the **Warrior University** (coming soon), and bring comfort and care to those in the hospital, in-person, or online. It's a small sacrifice for our 'lil Warriors on the front-line.

Please join Dr. Gary Lask, our Health-care Team Legacy Leader, and our



BREAKING NEWS

DOUBLE YOUR DONATION NOW. AND DOUBLE YOUR OUTREACH.

[READ MORE](#)

⇩

EXTRA! EXTRA! This just in from the wire: **We ALL have been once again blessed to be given an opportunity to double the donation!** We have an anonymous donor from CA who will match and donate **up to \$65,000** for research, care and support! Let's jump on this and really support their commitment to supporting each and every one of you in your journey with SWS, birthmarks and KT. NOW is the time and YOU can join in the fun of knowing we have UNITED for change and hope for today AND tomorrow!

volunteers as we aim for **1,000 caring individuals to donate a recurring \$10 a month** or more to provide a great impact for a lifetime! 🍌

COMMUNITY LEGACY LEADER: SUZANNE LITTLE

A Gift of Thanks to SWF Legacy Leaders

As a Legacy Leader, you will receive a beautiful acorn keepsake that symbolizes the lasting impact you're creating for SWS caregivers, patients, doctors, and researchers. By *planting seeds for a more fruitful tomorrow* you are contributing to the growth of a robust and resilient community, where challenges only make us stronger, like trees standing tall against powerful winds.

ROSE GOLD CHARM: Rose Legacy Leader, donate \$250

SILVER CHARM: Silver Legacy Leader, donate \$500

GOLD CHARM: Gold Legacy Leader, donate \$1,000+

Choice of: Charm (add to a charm bracelet), necklace, or lapel pin.





Around the World SWF International

In the age of thirty-one I have always committed to success by standing strong. These words were told to me when I was an elementary school pupil, “the stronger the winds, the tougher trees” which I do remember when confronting conflicts. My dad and mom, I see they have done so much for me to overcome those times “electricity discharge” in my brain not only taking me to the physicians in Turkey but also in the United States.

Keeping in a good mood during my school time was the key to make me able to earning high school degree through. I had never been humiliated by the classmates, teachers, and others because of my port-wine stain on my face (it’s just around eyebrow zone). Even during seizures I’d experienced on the grounds in the school, surely could not know what happened when unconscious, in the wake friends or the others given hands compassionately.

SWS is not known so much in Turkey. During inadvertent seizures outside our premises I would encounter stereotyping epileptic

attacks. Indeed, the attacks resemble epileptic ones but not because of suffering from epilepsy. I was told the seizures were epileptoid. That means my seizures exhibited symptoms resembling those of epilepsy but was not epilepsy.

I earned my twelve grade in Istanbul and then, we moved to a neighboring small city. As my mom is a licensed pharmacist, we set up our modest pharmacy and I’ve been working part-time there. I enjoy photography and playing PS games. All that I know about SWF comes from my dad who tells me all the time.

I spend time to improve my English and look forward to meeting the other specialist at an event in USA organized by SWF in the near future.

I hope that the SWS scientists will bring innovations that make our lives easier. 🍀

All the best,

Mert-Tekirdag
Turkey

Continued from page 6 Research Updates...

We heard encouraging new research exploring the impact of **GNMQ mutation on neurovascular** coupling. Unraveling this connection may deepen our understanding of the syndrome’s neurological aspects, offering potential avenues for targeted interventions.

As we navigate the path ahead, we are not only expanding our **Brain Vascular Malformation Consortium (BVMC)** project (read more on page 4) but also enhancing its impact. The recruitment efforts are ongoing, and we encourage everyone to contribute

to this transformational initiative. The “big data” from this project, combined with “big insights” from the SWFIRN researchers will make the journey ahead promising. With clinical trials on the horizon, your continued participation in our studies is invaluable, and together, we can make a lasting difference in the lives of those affected by Sturge-Weber Syndrome.

Thank you for your unwavering support, and here’s to another year of progress. 🍀

Board of Directors Update

It has been a delight and pleasure to have **Steve Emmons** serve as Treasurer the last three years. His cheerful demeanor and attention to detail have made the financial operations run so smoothly. We have had successive clean audits thanks to his leadership with our accounting team lead by **Jim Gilbert, CPA, with Jeremy Saavedra, and Liana Morse**. When you take a look back at the first annual report and see how far we have come from a little over \$2,000 to almost half a million dollars — look how much we all have accomplished! The SWF has been very blessed to have had outstanding financial leadership and there always seems to be another one to step in and carry on the example that **Pete Ober and Mitch Packler** set in the early years.



So, while sad to see Steve go we know where he lives! Jim Gilbert and the search team have found another terrific new Treasurer in **Dan Schmidt, CPA**. Jim and

Dan have known each other through Denver metro associations and Dan has consented to be nominated and approved for a two-year term. We are also fortunate that Dan has experience in the rare disease world working with organizations like Niemann-Pick. He lives in Colorado Springs with his wife and children.

John Rauschuber, a dedicated 30-year public high school teacher, and his wife Shelly, a second-grade teacher, have faced unique challenges raising their son Aden, born with Sturge-Weber in 1998. Aden's journey, marked by hurdles from birth through grade school, high school, and college, has been navigated with the unwavering support of the Sturge-Weber Foundation.

As Aden embarks on the search for employment, the Sturge-Weber Foundation continues to be a crucial source of support, connecting him with other young adults



who share similar life experiences. The foundation plays a pivotal role in fostering a sense of community and understanding among individuals facing the complexities of Sturge-

Weber syndrome.

In addition to his role as a devoted father, John has contributed significantly to education, teaching A.P. Government, Economics, Constitutional Law, and Philosophy. His commitment extends beyond the classroom, as he has served on various school boards, advisory boards, and dozens of educational committees throughout his illustrious career.

John Rauschuber has volunteered to serve on the Sturge Weber Foundation board with a clear awareness of the organization's evolving role for future generations. His decision to join the board is rooted in a strong commitment to ensuring the foundation's ongoing success and its ability to serve the needs of individuals affected by Sturge-Weber syndrome in the years to come. By dedicating his time, expertise, and efforts to the board, John aims not only to support current initiatives but also to play a pivotal role in transitioning responsibilities to the next generation. His involvement reflects a deep passion for making a lasting impact and underscores his dedication to seeing the foundation thrive as a sustained source of support, awareness, and assistance for future generations facing the challenges of Sturge-Weber syndrome.

Residing in West Los Angeles, the Rauschuber family includes two additional children, Weston (21) and Sarah (17). The Rauschubers exemplify resilience, compassion, and a dedication to both family and education. 🍎

Epilepsy Awareness Day at Disneyland



Epilepsy Services of NJ sponsored the Terrell Family to go to Epilepsy Awareness Day of Disneyland. Marissa and Julia arrived in CA and set up a table to represent The

Sturge-Weber Foundation. Two days of learning everything epilepsy. They had many talks about diagnosis, surgery, and so much more. We also got to work directly with our partners of Epilepsy Alliance America and Epilepsy Services of NJ. We ran into our sponsor Lara Falcon from

UCB and got some quality time with the seizure dogs. We also got to see Adrian too while we were there. There was a costume party pm Halloween and on the third day we all attended a day at Disneyland.

The takeaway there is nothing more important for time spent with people who understand what we go through and to learn from each other. The three days were truly a wonderful adventure and we are so thankful for our partners.

SWS Gift Ideas

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OFFICIAL SWF SHOP

SHOP TEESPRING

Two Shops

Zazzle
GIFT SHOP

SHOP ZAZZLE

SAVE 10% ON TEESPRING WITH "LOVESWF" • EVERYDAY SALES ON ZAZZLE



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UN**ITED**

Your donation is driving change

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- In **HONOR** of: _____
- Endowment Donation
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- I have enclosed a check
- Please pay by credit card* VISA MasterCard Amex Discover

Name on Card: _____

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**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: _____

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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 this form and mail to:

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



<https://sturge-weber.org/donate-today.html>