October 2025







## Community, Collaboration, Change: Our Strength Is in Each Other

by Founder and CEO - Karen Ball

The recent CCN/SWFIRN conference in Madison, Wisconsin was a very uplifting and unifying meeting. The attendees brought each of their levels of expertise in clinical and basic science research and gave us all updates and inspired us to do more and do better. The concurrent education summit, attended by some first time families and longtime supporters, was once again validation that the Foundation's ability to bring a wide Net of people from diverse walks of life is crucial to generating impact.

The meeting also drove home to me personally that wherever we are in this journey with Sturge-Weber syndrome and Port Wine birthmarks, we are each other's best teachers and guides.

United we inspire one another and ignite a plethora of great ideas to impact our daily living, our approach to learning more clinically/natural history and increasing collaborations to drive research studies.

I'm grateful to all who had input, attended and of course to Julia Terrell for her administrative and hostess with the mostess flair!

Never forget! You do NOT walk this journey with SWS alone. The SWF has created literally and figuratively an army of soldiers to march and defend you as well as shoulder to shoulder with you as you battle, each new wave of medical psychosocial and financial challenges.

Remember: whether for a reason, a season, or a lifetime you are always on our mind and in our hearts. As long as we're able, we will be here for each and every one of you that needs support, knowledge and hope.

P.S. Don't forget those Halloween photos you know I LOVE seeing them!

With faith, hope, and love,

CCN/SWFIRN & **Mini-Summit** 

by the





**7 Clinical Care Networks** 20 Clinical Docs & CRC 7 Researchers **2 Medical Students** 

A special thank you to Lisa Arkin, Dermatology; Yasmin Bradfield, Ophthalmology; and Stephen Falchek, Neurology for presenting.



#### **SWFIRN & CCN CONFERENCE**

**AND IN-PERSON MINI-SUMMIT** 

## United for Hope: Families, Researchers, and Clinicians Ignite Progress at SWF Conference

by Dave Holecek, SWS Dad

It was inspiring to be surrounded by families, researchers, and medical professionals this past weekend in Madison, Wisconsin, at the SWF Research Network Conference.

The Sturge-Weber Foundation has been a bright light for countless families over the years, offering both focus and hope. The Research Network meeting took that inspiration to the next level, uniting world-class researchers, clinicians, individuals with SWS and their families around a shared passion to accelerate clinical progress.

One of the most moving sessions came from two mothers, both nurses, who spoke with incredible strength about their daughters' hemispherectomies. Tears filled the room as they shared their struggles and their daughters' remarkable perseverance... only to be replaced with pure joy later that evening as those same girls ran through the conference space, laughing and playing with one another and their siblings.

The research updates were equally inspiring. The presentation of the SWF Natural History Study (an integrative database of medical records, images, and serum markers) was extraordinary. Dr. Loeb and his team have built something truly unique in rare disease research (I am a biotech/ rare disease executive), creating a resource that will directly improve clinical trial design and accelerate the timeline for future disease-modifying therapies. I look forward to the publication of more detailed results. The dynamic nature of the condition, along with the progression of symptoms, resonated strongly with my own family's experience. The brain's ability to remodel itself is remarkable and is always be a source of hope.

Collaboration was another powerful theme. In breakout sessions with the University of Wisconsin medical team, I saw not only academic brilliance but also deep compassion for families living with SWS. Conversations over dinner with physicians and PhD scientists (including some studying the GNAQ mutation and zebrafish models) reinforced this commitment. Many attended at their own expense simply because of their passion to advance SWS knowledge.

The weekend reminded me that we are not alone. Alongside the challenges of seizures, headaches, cognitive and behavioral issues, and uncertainty, there exists a committed global community of families, physicians, and scientists working together toward better outcomes. I left with renewed hope for the future of our children and families — and with deep gratitude to Karen and Julia for their tireless dedication.

## What attendees had to say...

"Attending the recent Sturge-Weber scientific conference was an incredibly positive and inspiring experience. It was truly energizing to be among such a brilliant and dedicated group of doctors, scientists, and researchers—all working together with a shared purpose: to uncover answers and drive meaningful progress. The collaboration gave me renewed hope and a deep sense of gratitude. Seeing this passionate community unite around advancing care and understanding of Sturge-Weber syndrome reminds us that we are not alone—and that real breakthroughs are within reach."

-LINDY





# Unlocking the Future of SWS: BVMC Project Reveals Groundbreaking Insights for Patients and Families!

by Jeff Loeb, MD, PhD





The BVMC project has come to fruition! At the CCN/SWIFRN meeting in Madison, Drs. Jeffrey Loeb, Dilip Pandey, and Biswajit Maharathi from the University of Illinois at Chicago gave us a preview of much to come. The BVMC project that built the most comprehensive natural history study on Sturge-Weber syndrome with high resolution data on over 140 patients. This includes clinical stories,

medication treatments and brain imaging and covers neurology, dermatology, and ophthalmology.

The studies are revealing remarkable changes in the brain over time that coincide with important clinical changes. Dr. Csaba Juhasz from Wayne State University reviewed hundreds of brain MRI studies and shows some remarkable fluctuations in findings and changes over time that are being linked to neurological symptoms.

Dr. Biswajit Maharathi also showed how the clinical dashboard developed from this project is being repurposed into a personalized electronic health record to be given back to patients, their families, and their doctors. This had led to the formation of a Technology Advisory Board at the foundation to oversee and raise funding and awareness for this initiative that could transform clinical care and research for SWS patients. During the 'data blast' the latest findings in dermatology (including laser treatments), ophthalmology (including glaucoma), and neurology.

Dr. Pandey has put together the largest series on stroke like episodes, defining what they are, how they occur, and what we can do to stop them.

Finally, Dr. Loeb ended the conference with a heart felt discussion on the cognitive, behavioral, and emotional aspects of SWS. He noted that the changes that occur in the brain alone, including progressive calcifications, could drive many of these mental health changes and suggested a new therapeutic that could greatly improve things.



THANK YOU TO OUR
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# Congrats!

**Aimee Luat,** MD was promoted to Division Chief (Pediatric Neurology, Epilepsy) at the Children's Hospital of Michigan.

**Anna Pinto**, MD, PhD is moving to Emory University to head up the new SWS Department.

John McLaren, MD will be the new head of SWS clinic and replacing Anna Pinto at Boston Children's Hospital.



Please join us to celebrate the retirement of Dan Miles, MD is retiring from NYU Langone.

Stephen Falchek, MD is retiring from Nemours Children's Health

# Language Matters, Lives Matter: How Collaboration Drives Sturge-Weber Solutions

Dr. Lisa Arkin, Dermatology - CCN University of Wisconsin-Madison



The 2025 Sturge Weber Clinical Care and Research Network Meeting in Madison brought together an extraordinary group of clinicians and scientists dedicated to advancing care for patients with Sturge-Weber syndrome. Experts in dermatology, neurology, neuroradiology, oncology, and ophthalmology joined to share insights across disciplines. Keynote speaker Dr. Kris Saha (UW-

Madison) presented "Advances in Cell and Gene Therapy Using CRISPR Genome Editing: From Reading to Writing the Genome," highlighting how cutting-edge genetic technologies may shape future therapies for rare diseases.

Highlights included review of new imaging data to improve diagnosis of SWS, a review of Brain Vascular Malformation Consortium findings, and plans for the next phase of analysis. Importantly, the group emphasized that language matters—what we call things determines how we treat them. A patient panel amplified the power of engagement, helping to shape priorities in optimizing outcomes for the skin, the brain and the eye. This year's meeting reinforced the power of collaboration and shared vision in driving progress for the Sturge-Weber community.

There was discussion on how studies performed in collaboration with the Foundation will set the stage for new clinical trials and ways to improve clinical care. All participants agreed on the importance of addressing the emotional impact of the disease and advocated for longitudinal testing of neuropsychological measures as important outcomes.







## What medical students had to say...

"We had the privilege of attending the 2025 CCN/SWFIRN Science Conference, and it was truly an invaluable experience.

As medical students, we learned so much from dermatologists, neurologists, and other clinicians/scientists in the field, as well as from families who generously shared their journeys with Sturge-Weber syndrome.

Being part of this conference gave us a deeper understanding of both the medical complexities and the human impact of Sturge-Weber syndrome. We are grateful to the Sturge-Weber Foundation for welcoming us into this inspiring community, and we look forward to carrying these lessons with us as we grow into our future roles as physicians."

#### -Nicole Chin

Medical Student at Loyola University

#### -Keana Khodadad

Medical Student at Rosalind Franklin University



#### Give Your Child a Brighter Future by **Advancing Sturge-Weber Research** with a Simple, Painless MRI

by Csaba Juhasz, MD, PhD



The NIH-funded, prospective study on advanced neuroimaging in Sturge-Weber syndrome (SWS) is still recruiting participants before the current phase of the study is expected to end in February, 2026. The study is performed by a group of investigators at Wayne State University (in Detroit, MI), led by Dr. Csaba Juhasz, professor of Pediatrics and Neurology. The team also includes Dr. Aimee Luat,

chief of Pediatric Neurology at the Children's Hospital of Michigan, Michael Behen, PhD, neuro-psychologist, as well as MRI experts. Study procedures include a non-invasive brain MRI and a neuropsychology evaluation, performed in young patients (<28 years of age) with the clinical diagnosis of SWS.

The study, that started back in 2003, has recruited more than 100 patients, partly with the assistance of the Sturge-Weber Foundation; many patients have been followed for years. The study findings have led to a better understanding of the changes in brain blood vessels and tissue abnormalities during the early disease course of SWS, and how these can lead to progression and/or stabilization of clinical symptoms. The study also introduced advanced new imaging technologies in SWS diagnosis, including the use of susceptibility-weighted imaging (SWI), that provides a unique insight into key brain abnormalities such as abnormal deep veins and progressive calcifications. The imaging findings also shed new light on the mechanisms of epilepsy and processes of brain plasticity as well as vascular remodeling. The study results have been published in major medical journals. At the end, the study findings are expected to yield key imaging markers that will be used clinically for diagnosis, prognostication, and, hopefully to assist new treatments in future clinical trials.





Learn more about our Memorial Garden here.

#### **SUPPORT OUR MISSION TODAY!**



#### THE STURGE-WEBER FOUNDATION

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#### October 4, 2025 @ 12:00 pm PST Women's Health Virtual Mini-Summit

- Explore why multidisciplinary clinics for young women with epilepsy and Catamenial Epilepsy with Dr. Christina Briscoe
- · Hormones throughout the life cycle with Dr. DiVasta Thank you to the team at Boston Children's Hospital. This will be an exciting talk. **REGISTER HERE**

Sponsored by:



#### October 15, 2025 @ 7:30 pm EST **Mental Health Chat for Caregivers and Parents**

These virtual chats are an opportunity to meet other caregivers and parents of SWS, KTS, and PWB children and adults. Moderated by Luis Sandoval, PhD, SWF Task Force Leader for Mental Health. This is an opportunity to learn, laugh, and support each other on this caregiving journey.

#### **REGISTER HERE**

#### October 8, 2025 @ 12:00 pm PST Mental Health Chat for Patients 21+

Our Mental Health virtual chats allow one to meet other adults (21+) with SWS, KTS, and PWB. Register below, and we will send you the Zoom link on the day of the event. Moderated by Luis Sandoval, PhD, SWF Task Force Leader for Mental Health. This is a great opportunity to bond, laugh, and chat about any mental health or general life issues you may be experiencing in a supportive space. We hope to see you there! REGISTER HERE

Continued on next page...

### **Member Photos**



This is baby Noah, and he was just diagnosed with Sturge-Weber syndrome in August. Noah has a very light birthmark that practically vanishes when he's feeling happy and playful. When he cries or strains, it darkens, and that is the clue that helped his parents advocate for a diagnosis. He has confirmed brain involvement, and started his medications this week! When in doubt, seek out the experts. We are so grateful for the experts, this foundation, and the SWS family community!







October 20, 2025 @ 12:00 pm PST SWF Be The Torch, Be The Light Join Ansley and Donald for a night of Hope and all things religion. REGISTER HERE



November 6, 2025 Virtual Mini Summit with UC Irvine @ 4:00pm-5:30pm PST REGISTER HERE

















# SURVEY SAYS: Community Voices Highlight Ongoing Needs at SWS Family Feud Game Night

During the dinner after the mini-summit, we hosted a Sturge-Weber Family Feud Game Night with patients, caregivers, and doctors and clinicians. This ice breaker game was more than just for laughs, it had a powerful purpose too.

Through a series of survey-style questions, participants shared their honest experiences with Sturge-Weber syndrome (SWS), shedding light on the real-life challenges the community continues to face.

We asked 24 patients: "What is the most important impact of Sturge-Weber on you or your family life?" Top responses included:

- Self-Acceptance: Many shared struggles with not fitting in, mental health challenges, and the journey toward self-love.
- Lack of Doctor Knowledge: Patients expressed frustration with the limited understanding of SWS among healthcare professionals.
- Migraines and Headaches: A common and often debilitating symptom for many.

Next, we asked 24 caregivers: "What's the most concerning part of caring for someone with Sturge-Weber syndrome?"

The most common concerns were:

- Seizure Control and Medication Management
- Uncertainty About the Future of their child or loved one
- Finding Knowledgeable Doctors who truly understand SWS and its complexities

Finally, 48 patients and caregivers responded to: "What's one area of Sturge-Weber co-morbidities you wish you knew more about?"

The top areas identified were:

- Need for More Research Across the Board
- Understanding of Learning Disabilities and Mental Health
- Access to Affordable Insurance and Resources

WE ASKED 24 PATIENTS: WHAT IS THE MOST IMPORTANT IMPACT OF STURGE-WEBER ON YOU, OR YOUR FAMILY LIFE?

Self-Acceptance (Not Fitting In to Society's Appearance, Self-Love, Mental Health Issues)	10
Lack of Doctor Knowledge	5
Migraines/Headaches	1

WE ASKED 24 SWS CAREGIVERS: WHAT'S THE MOST CONCERNING PART OF CARING FOR SOMEONE WITH STURGE-WEBER SYNDROME?

Seizure Control / Medication For Seizures	9
What will happen to my child in the future	6
Finding a doctor who understands all the needs of SWS	4

WE ASKED 48 PATIENTS AND CAREGIVERS: WHAT'S ONE AREA OF STURGE-WEBER CO-MORBIDITIES YOU WISH YOU KNEW MORE ABOUT?

More Research All Over	19
Learning Diabilities/Mental Health Issues	5
Affordable Insurance/Resources	3

These responses clearly show that while progress has been made, the SWS community still faces significant gaps in medical knowledge, mental health support, and access to comprehensive care.

The night was full of laughter and camaraderie—but the messages shared were powerful.

Together, we're pushing for better research, better care, and a better future for everyone impacted by Sturge-Weber syndrome.

# TEAM BRIELLE Rhode Island Walk September 20



Listen to the podcast appearance on WPRI 12+ with the Coutu family and Karen <u>here.</u>























