A PUBLICATION OF THE STURGE-WEBER FOUNDATION

Branching Out

DECEMBER 2017

ROOTS TO RESEARCH

ARE YOU A DISCERNING MEDICAL CONSUMER?

GROWING GOLDEN: SWS ADULT SERIES

The Sturge-Weber Foundation
MAGAZINE

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Branching Out

December 2017

IN THIS ISSUE . . .

On the Front Cover: Dana Davis and son, Bryce.
On the Back Cover: Amelia Zinski, daughter of Hayley Zinski, water sking with family friend. Beau Arch sports his Halloween costume before the big day.

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).

Klippel-Trenaunay syndrome, or KT, occurs as the result of a congential vascular malformation in an extremity, such as an arm, leg or foot.

The SWF is a clearinghouse of information for Port Wine Birthmarks, Sturge-Weber syndrome and Klippel-Trenaunay syndrome.

THE 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, SWS and KT.

As a clearinghouse of information, the Foundation will seek information regarding management and treatment techniques and suggestions concerning aducation and emotional support and will facilitate the dissemination of appropriate information.

If, in facilitating research on PWB, SWS and KT, the Foundation provides financial or other support to a particular research project, the Foundation will base its decision upon need, the Foundation's financial resources and medical advice.

The SWF is a member of the National Organization for Rare Disorders (NORD), American Brain Coalition (ABC), The Coalistion of Skin Diseases (CSD), and the Association for Research in Vision and Ophthalmology (ARVO).

CONNECTING WITH KAREN

What a great start to the 2017-2018 fiscal year!

The first quarter of the SWF's fiscal year was a whirlwind of activity, collaborations and connections!

The July Conference in Cincinnati was fun for one and all with a great mix of insightful lectures and moments to just hang out and get to know one another or reconnect. The Clinical Care Network and Dr. Adrienne Hammill truly did a stellar job coordinating with Julia Terrell in pulling it all together . . . thank you!

The Falmouth Road Race in August was a phenomenal success and the dedicated runners and sponsors raised a record number of crucial donations that created direct impact for patient and family support and research endeavors. Pam McIntyre, Board Chair and Jessica Melo have dedicated themselves tirelessly to ensure each year brings great rewards. I think THEIR greatest rewards are yet to come!

The Clinical Care Network (CCN) and Patient Engagement Network (PEN) meeting in October, led by Dr. Jeffrey Loeb, SWF Chief Clinical Strategist and Brian Fisher, VP Operations and Corporate Partnerships was a fabulous example of collaboration and focus. Each specialty set goals and action items from a list of identified gaps in our knowledge or delivery of care in each speciality. Susan Finnell and Julia Terrell made sure everything flowed smoothly and I DO appreciate all they did to chart a new course and increase the pace of discovery.

As the season of thanks and giving is upon us, I want to wish all of you the very best of cherished memories and my thanks for thinking of the SWF in your holiday giving . . . for a reason, a season, or a lifetime! Your gifts of time, talent and treasure help maintain a solid foundation to continue to build upon our earlier accomplishments and creat new ones too . . . I believe and I hope you will too!

Merry Christmas, Happy Hanukkah, Happy Kwanza and a Happy New Year!

With faith, hope and love,

Karen

ARE YOU A DISCERNING MEDICAL CONSUMER?

FROM KAREN BALL'S BLOG

> *It's pretty much standard, after receiving any diagnosis today, individuals take to the internet to delve further in their understanding.*

Lately, due to a series of events, I have been ruminating on the plethora of online and traditional resources at our fingertips. It's pretty much standard that after receiving any diagnosis today individuals take to the internet to delve further in their understanding of their diagnosis. They rely on their healthcare provider to guide them in the dispensation of their healthcare, diagnostic needs and follow-up care. It is our responsibility as a discerning medical consumer to have due diligence in all matters relating to our own or our loved ones health. We have a finite amount of time and money and with proper awareness and planning we can make the best use of those precious commodities.

The Sturge-Weber Foundation will be developing a series of articles and resources to assist you in becoming a discerning medical consumer. The series will address ethics and the doctor-patient relationship, medical necessity, communication, medical records and record keeping, support and resources and much more in general and also as it relates specifically to a Sturge-Weber diagnosis. We will be engaging your participation and input throughout the series with a variety of polls and surveys. Some of the best advice I know comes from those who have gone before you but one must also temper that advice and advice from healthcare providers as it pertains to your own unique circumstances…be a discerning medical consumer.

A Sturge-Weber diagnosis initially hits you in the gut like a sucker punch! Once you can catch your breath most of you are off and running to find THE best doctor, THE best resources and THE best support you can amass. The SWF has been a respectable and comprehensive network of learning and research to help you confidently launch this new phase of your life. We can assist you to ascertain what needs you have and establishing a course of action.

Of course, then you move on to the "veteran" stage. You know the one where the initial crisis has passed and you learned to cope and exhale instead of holding your breath. The SWF wants to be sure you don't live in angst but never forget SWS is a lifelong condition that has evolving medical and psychosocial needs that must be addressed. This series is equally important for you! We will be addressing many issues related to aging with SWS in the coming year and as a discerning medical consumer "veteran" we will be engaging your wisdom and addressing your evolving and unique needs as well.

Remember, We Are Always Better Together!

ON THE WEB | IN THE NEWS

www.sturge-weber.org

THANKS TO DAN DORNEY

SWF thanks Dan Dorney for serving on the Board of Directors and as Treasurer. Dan's service has been steadfast and his commitment to the SWF has been greatly appreciated. Board service is not onerous, but is an elevated level of commitment and he guided the SWF financially leaving us in great stead.

ONLINE SHOPPING

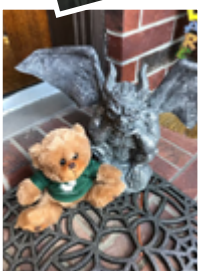
The SWF Online Store is up and running! Check it out at *http://sturge-weber.org/who-we-are/swf-online-store*. Currently we have SWF wristbands, backpacks, canvas tote bags and of course, Webster, the SWF Road Warrior bear. Route to a Cure t-shirts will be available soon, so check back often!

WEBSTER IS ON THE ROAD!

Meet Webster, SWF's roving road warrior bear. Webster comes to the Foundation to provide a window into the lives of patients and families that live daily with Sturge-Weber syndrome and other Port Wine Birthmark conditions. As you know, these conditions last a lifetime and are unique to each individual. Though some of the symptoms are similar, no patient experiences the disease quite the same.

Webster tells their story in pictures as he travels along with them. There are good days and bad days. There are victories and setbacks. Webster shares it all in an effort to impact the public and heighten awareness across the globe.

Would you like to have Webster share your story? Perhaps you would like to send Webster to a patient to encourage them along the way? Check out Webster and Cody Marshall's journey to the 2017 International Conference in July in Cincinnati, OH at *https://vimeo.com/237774886*. **YES, it really IS that much fun!**



Photos top to bottom: Brielle Coutu, Marissa Terrell, Webster at the BVMC, Houston Airport, Halloween in Houston, TX.

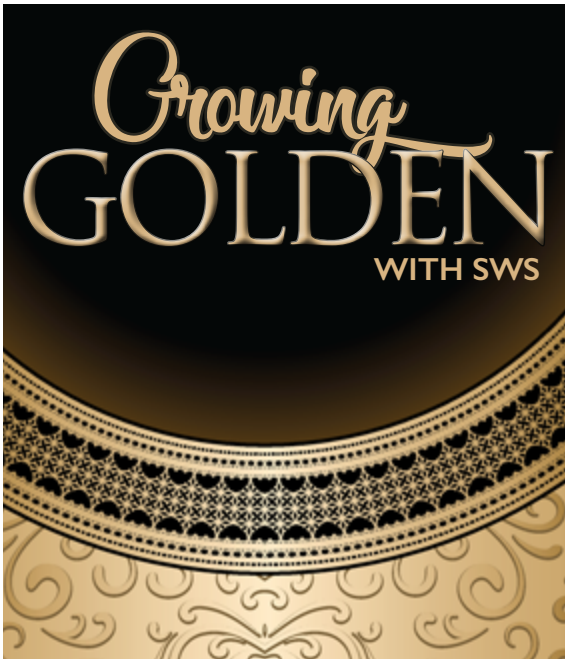

THE STURGE-WEBER FOUNDATION
Reunion of CHAMPIONS
benefitting patients with SWS
and continuing critical research
The Main Event
Friday, February 16, 2018
The U. S. Grant Hotel
Bivouac Ballroom
326 Broadway, San Diego, CA 92101
619.232.3121
Cocktails | 6:30 - 7:30 PM
Hor d'hoeuvre Buffet | 7:30 - 9:00 PM
Special Entertainment
Attire: Business or Cocktail Dress
HONOREES
Richard Felten, M.S.
FDA Office of
Device Evaluation
Dr. Michael Gold,
M.D., FAAD
Gold Skin Care Center
Nashville, TN
For details and registration:
http://sturge-weber.org/
for-professionals/

SPECIAL SERIES

Growing GOLDEN WITH SWS

Sturge Weber syndrome in all its manifestations is a lifelong challenge. At birth, parents are alarmed and afraid, which impels them to find answers and seek medical care. Childhood brings constant medical activities that become almost routine, but with the caution that anything might happen. Teen years bring changes both in SWS-centered care and typical growing-up medical and social/emotional events. The step into adulthood has challenges that transcend just the medical aspects.

Mature adulthood brings its own set of questions.



For the independent person, the juggling act with work, medical appointments, social responsibilities can test his or her fortitude and inventiveness. "Can I find a knowledgeable doctor to be with me on this lifelong journey, or do I have to become the tutor for every new physician I encounter?"

For the parents of a dependent or semi-dependent adult, in addition to continuing to monitor the health of the adult child, there are concerns about "handing over" and "letting go". Today's society has numerous opportunities for adults with disabilities to experience independence that they can handle, without the risk of being cut off from the assistance and support of families. There are ethical and legal situations to be faced.

The SWF has a valuable resource as we contemplate hopefully growing older - we have our adult members who have endured and experienced SWS/PWB/KT and are willing to share their hard-won wisdom.

We are embarking on a project to involve adults with SWS. Please share with us questions that have puzzled you or questions that just emerged. We will consult the doctors who can assist and provide some answers. There will be an "Ask the Expert" section in each issue in the future, geared to "grown-up" questions, not pediatric.

There are also many parents who have a different perspective depending on the independence and relationship with their adult child. Their stories can provide signposts.

COMMON QUESTIONS

These are questions that have been posed to us by a group of adults with SWS or parents of adults. It is not a complete list and you are invited to send us any other concerns that occur to you. *We will save the social/emotional and financial/insurance/employment related questions for another issue.*

Please be aware only some of these questions may or may not apply to you. Some may have no connection to SWS, PWB or KT. Any of these questions should be brought to the attention of your own doctor. The SWF can only offer very general information and cannot give medical advice. The SWF can assist with providing information on specialists in all areas that might be someone you can investigate. No one doctor can answer all questions about SWS, PWB or KT since they are multi-system disorders. Additionally, a specialist who is familiar with your specific question would have to see you as a patient before making a recommendation.

Neurology
- Are AED medications more or less effective as you get older?
- Should I get an MRI at my age, (not a child) or is it not very helpful?
- What would trigger a seizure in an adult who has never had one? Is it related to SWS?

Dermatology
- What should I do to prevent the natural drying of my skin that comes with aging? Does laser treatment dry my skin?
- What are the proven methods to treat adult PWB? How do you reduce hypertrophy and/or nodules (blebs)?
- Can laser treatment be effective as you get older? What is the best post-laser skincare regimen to reduce ulceration, scabbing /scarring?
- Would it be healthier for my skin NOT to use makeup? How high SPF should I use on sunblock?
- At what age or stage is it too late to perform laser surgery on a birthmark? For older patients who begin to experience thickening, is there still benefit in continuing surgery?
- Is it likely that hypertrophy will increase in middle age? How and why?

Ophthalmology
- I do not have glaucoma, but my vision seems to be getting worse. Is this typical for my age, or does SWS have something to do with it.
- Can I have Lasik surgery on my non-SWS eye(s).
- Can I have cataract surgery or would it be risky?
- What should be the target IOP for people with SWS related glaucoma?
- What about medications like antihistamines, steroids,) and do they affect glaucoma?

Endocrinology
- How does my hormonal level affect my SWS? How does SWS affect my prostate health? GYN health?
- Is there a connection between diabetes and any other endocrinology concern that my SWS causes?

Klippel-Trenaunay
- Is surgery the only option for varicose veins? Is sclerotherapy indicated and is it successful?
- How effective are compression garments?
- Have lasers been successful in treating KT?
- What are typical situations KT patients anticipate as they age? Is the vascular surgeon the proper clinician to treat KT?
-

Below are questions on adult concerns by condition.

High Blood Pressure
- At what age should I begin to monitor and treat hypertension (high blood pressure)? Who should be doing this? Internist? Family Practice?
- What blood pressure medications are OK for people with SWS/PWB? If I also take AEDs?
- What effect, if any, does SWS or PWB have on hypertension? What about glaucoma? Or the other way around, what effect does hypertension have on port wine birthmarks, SWS, or glaucoma?

High Cholesterol
- At what age should I begin to monitor and treat high cholesterol? Who should do this? Internist? Family Practice?
- What cholesterol medications are OK for people with SWS, PWB? What about statins? If I also take AEDs?
- What effect, if any, does SWS or PWB have on my cholesterol level. Do my cholesterol levels have any effect on SWS or PWB?

Arthritis
- Does SWS have any connection to joint stiffness or pain? What about RA? (rheumatoid arthritis?)
- What about prescription meds for arthritis – some can be powerful. If I also take AEDs?
- How about over-the-counter remedies and supplements?
- Does SWS put me at higher risk for osteoporosis?

Diabetes
- When should I begin to monitor my blood sugar levels. What lifestyle changes should I make to prevent diabetes?
- What about diabetes medications? If I take AEDs?
- Autoimmune disorders
- How does having SWS or PWB impact a disorder like RA (rheumatoid arthritis) or MS (multiple sclerosis)?

Dental
- What is the risk of dental loss/periodontal disease because of long term use of oral medications commonly used by those with SWS (anti-seizure or glaucoma meds).
- How risky is tooth extraction and what precautions should dentists take to prevent excessive bleeding?

General
- Should I have flu shots? Shingles vaccine?
- Is weight gain connected with SWS? Can I diet safely?
- Do I have to add more specialists to my list of doctors as I get older? Cardiology, rheumatology, nephrology? Urology?
- If my neurologist has been the main SWS clinician, should he/she be the one monitoring general health concerns? How do I keep all the areas of healthcare straight so that I am not overlooking something?

REAL LIFE SUGGESTIONS

Theresa Lamb of Boston shares some thoughts: Thanks for asking about blood pressure. For me, my bloopressure changed dramatically when I left a high pressure job (I have my current job now for 2 years - very little stress). I am also doing more things for myself such as being in a bowling league for 2 years, going to minor league baseball games. Shortly after I turned 40, I had my firs seizure and found the SWF. Since then my blood pressure has lowered even more because I have met many wonderful friends who understand.

Annette Blazek from MN adds:
I have had cataract surgery and there was no problem.

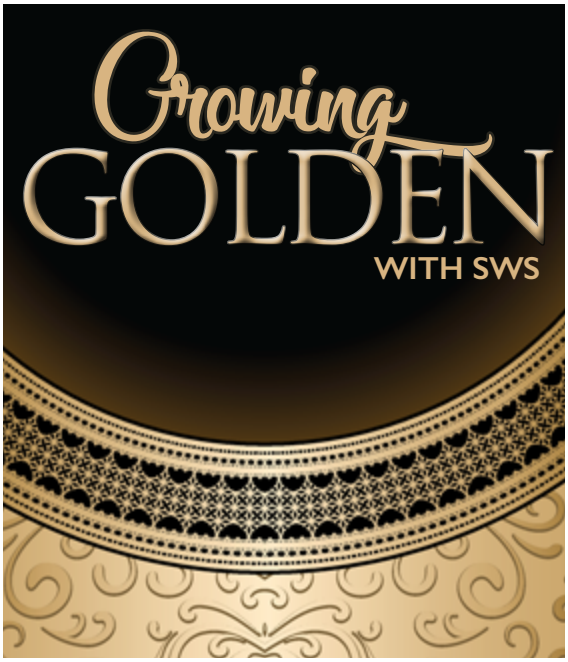
About weight gain, sometimes your anti-seizure meds are responsible for weight gain. I found that out when I changed to another med and realized that it wasn't just me "not being able to control what I ate"

Lakshmi Menon in India shares:
I am glad that women are keeping record of their health issues and body changes. We need to understand our bodies instead of relying completely on doctors for treatment and prescriptions. And sharing info about their conditions with SWS will certainly help others analyze their own conditions. I go to a nearby yoga center – for about 40 years intermittently – but because of my total knee joint replacement I am unable to sit cross legged on the floor (a typical yoga stance). So I sit on a chair and do what I can, mainly breathing exercises and meditation.

But I am taking drugs like methotrexate for the past 8 years and now also steroids for join inflammation and pain. I am always troubled about the quantity of medication I am taking and the impact on my health, alternating between anguish and guilt. No one wants to be in pain all the time.

Nina Weber, mother of adult daughter says:
They were advised that MRIs are only necessary when the patient displays new symptoms. Her daughter always has yearly Pap tests and mammograms, like any typical woman. They always make sure she has her flu shots as well as the shingles vaccine, since she had chicken pox as a child. She was also advised that there is a higher risk of osteoporosis since her daughter was on Phenobarbital.

Rebecca Szorcsik of New Jersey says:
I noticed that my birthmark started changing around 37ish. Had to restart the laser treatments. They said this is very typical as the birthmark ages.



ONE PARENT'S EXPERIECE

Margot and Richard Slater's daughter, Kim Slater, is an adult member of the SWF PEN (Patient Engagement Network). Margot shares her experiences and thoughts with us.

Up to now I have never really discussed with anyone about having a child with a 'birth defect", as it was explained to us for years. Certainly, not now having an adult child with a rare disease either. Our experience has always been that is was just assumed we would do what we needed to do for our child. Any ramifications it might have for us, or her then 2 1/2 year old brother, were just part of life.

Kim has always said that we were her advocates for a long time. I feel I learned to be an outspoken woman because of her. I learned how to get things done when I was told it couldn't be. I learned how to say "no". As a result, Kim has said that I provided her with a good role model for learning how to advocate for herself. It was difficult letting go after doing the job for so long.

I definitely have concerns for her as an independent adult. Why does it appear that there are not many other independent adults? Do I want that answer?

As a child Kim was pretty much asymptomatic except for the glaucoma and port wine stain. Now, I realize how very lucky we were (are). As a result, she was able to become independent. However, as an adult she has come down with various other maladies. Is the POTS related to SWS? Are the seizures SWS? Was she always destined to lose her eye? Will she develop other symptoms because she is an adult?

It also seems that fewer and fewer neurologists are truly informed about adults with SWS. Why can't pediatric neurologists continue with their patients since they seem to be the experts on SWS? I am concerned that there will be fewer experts as she gets older.

We went with the approach that she was to go out in the world just as anyone else. We made sure she participated in everything that everyone else was involved in. We wanted her to develop self-confidence as herself. To learn not to judge a book by its cover, so to speak. We didn't use make-up until she asked. We didn't want her to think we needed to cover her up to be out in the world. We didn't treat her differently from her brothers. We thought this was our normal and so was she.

I have concerns about mental health issues. Many questions. Will Kim be able to be an independent adult for the rest of her life? How do we handle the different issues that seem to develop as adult? We can't make an adult do things or make choices that we think will help. How do we help an adult with SWS? We will always have concerns, she will always be our daughter.

We are here . . .

for a reason,

a season,

a lifetime.

We are better together.

Thanks to the SWF families and adults who contributed to this report – so far. We welcome other comments, questions and stories. Thanks to Lakshmi Menon, Nina Weber, Annette Blezak, Jill Barnes.

TRANSITIONING TO ADULT CARE

The SWF has always tried to help families manage medical care for their adult children, but it is often answered on a piecemeal basis. The pediatric SWS team many families have assembled doesn't always dovetail with adult care needs.

Our friends and colleagues at the Genetic Alliance Community have shared some valuable insights and links to answer these concerns. Especially helpful is this focused list of concerns:

- Educational transition planning via IEPS, IFSPs and 504 plans seldom bring the aspect of health care transition into the process.
- Most Pediatric practices do not have a formal process in place for transition
- Adult primary care providers receive little training on working with persons with disabilities and complex care needs
- Families work to have a medical home/community for their children, but research shows most youth and their families are unprepared for transfer of care to an adult system
- Financial reimbursement for the work and activities surrounding health care transition remains challenging.

There are a quite a few national and state resources on special health care transition. Here are a few:

- Got Transition - http://www.gottransition.org
- Autism Speaks Transition Toolkit - http://www.autismspeaks.org/family-services/tool-kits/transition-toolkit
- New England Consortium Transition Toolkit - http://newengland-consortium.org/for-families/transitioning-teens-to-young-adults/transition-toolkit
- Florida HATS - http://www.floridahats.org

While these are not specific to SWS, they can be valuable tools for understanding the process, another "how to do it" list for parents and their soon to be adult kids.

A recent issue (April-May 2017) of Neurology Now magazine had some guidelines for parents. They recommending beginning the conversation about transition with their present neurologist when their child turns 13 years of age. Hopefully the search will be completed by the time their child reaches 18-21 years of age.

The Sturge-Weber Foundation has several Social Media platforms for patients and family members to join. We encourage you to check them out and start connecting with others!

For details, go to http://sturge-weber.org/participate/swf-facebook.html.

Sturge-Weber Syndrome

Klippel-Trenaunay

Port Wine Birthmark

Roots to a Cure

RESEARCH REPORT

CHARTING IMPACT - IN THIS TOGETHER!

This is an exciting time for the SWF and for all of you impacted by the diagnosis or a Port Wine Birthmark! We had a record number of applications for Lisa's Research Fellowship and for applications to become a Clinical Care Network center (CCN). The strategic direction for research and patient engagement is renewed and focused on manageable goals with most importantly action items and leadership.

We each bring our unique perspective and cultural diversity to share our collective wisdom to improve the quality of life and care for those living with these conditions. We are united and invigorated to do all we can within our cicles of influence to ensure this happens. A vital and key aspect of research is tissue donation. There is never a right time to make a delicate request for tissue donation . . . but the best time is before surgery occurs and hopefully one will never need to invoke the process!

There are some parents who just can't think of it either before, during or after their child's brain surgery . . . others are right there if it will help another family not endure what they did!

I implore you to share with researchers and your fellow members living with SWS the most precious gift of tissue, blood and knowledge which will have a lasting impact.

Look what it did for finding the GNAQ Mutation!

We always need to replace it and knowing BEFORE a surgery enables researchers in need of fresh tissue to ensure they can keep investigating leads.

You have heard of the proverbial hit by the bus saying? Well, if any one researcher/clinician goes down the SWF has built a comprehensive research (SWFIRN) and clinical (CCN) program to be sure the forward momentum and collaborations continue. Please join us in doing your part with your time, talent and treasure to support this amazing and energized program today!

In This Research Supplement:

and more!

REPORT: PeDRA CONFERENCE

The annual Pediatric Dermatology Research Alliance (PeDRA) meeting was held in Denver, CO and brought together leading pediatric dermatologists and young investigators, fellows, medical students and advocacy organizations. The PeDRA group has grown in influence and generating targeted investigations and studies which drive inroads to undeerstanding disease states, quality of life impact and genetic aspects. The unique approach of bringing together this diversity of experience and ideas with smaller breakout sessions creates lots of enthusiasm and brainstorming. The SWF was a conference sponsor and Karen Ball participated in the vascular birthmarks section. Look for surveys and call for participant communications in the near future. The CCN was represented by 7 centers around the country too!

Dr. Moise Levy-Dell Childrens and Dr. Ilona Frieden enjoying the PeDRA Conference.

Karen Ball participates in a seminar on patient advocacy at the PeDRA Conference.

REPORT: LISA'S FELLOWSHIP RESEARCH AWARD

Colette Bichsel, PhD of Boston Children's Hospital Receives 2017 Lisa's Sturge-Weber Foundation Research Fellowship Award

The Sturge-Weber Foundation named Colette Bichsel, PhD of Boston Children's Hospital the recipient of the 2nd Lisa's Sturge-Weber Research Fellowship Award.

Research is defined as "patient-oriented research" conducted with human subjects, or "translational research" specifically designed to develop treatments or further various areas of scientific discoveries within Sturge-Weber Syndrome (SWS). The funds for the Lisa's Research Fellowship Award are from a family that wishes to remain anonymous to focus the attention on "Lisa's Sturge-Weber Foundation Research Fellowship Award." Eight applications were received for this award.

Dr. Bichsel is a postdoctoral researcher at Boston Children's Hospital and Harvard Medical School. Her fascination for how tissues self-organize led her to the field of vascular research. Before joining Vascular Biology Program in 2016, she completed her PhD in Biomedical Sciences at the University of Bern, Switzerland. She holds a Bachelor's and Master's degree (summa cum laude) in Bioengineering from the Swiss Federal Institute of Technology. She uses this technical background to develop tools that are suitable to study how blood vessels assemble and function.

"The Sturge-Weber Foundation is honored to present Dr. Bishcel with the 2nd Lisa's Sturge-Weber Research Fellowship Award and look forward to the positive impact that she will make in the lives of the patients and families with SWS," stated Karen L. Ball, Founder and CEO.

For details on Dr. Bichsel's research, vist the SWF website at http://sturge-weber.org/for-professionals/lisas-research-fellowship.html.

COLLABORATION AND INTEGRITY: WE ARE BETTER TOGETHER

THE STURGE-WEBER FOUNDATION CLINICAL CARE NETWORK + THE STURGE-WEBER FOUNDATION RESEARCH NETWORK + THE STURGE-WEBER FOUNDATION PATIENT ENGAGEMENT NETWORK

REPORT: CHICAGO EDUCATION FORUM - LURIE CHILDREN"S HOSPITAL

The first Education Forum co-sponsored by the SWF and the Ann & Robert H. Lurie Children's Hospital of Chicago was held October 21, 2017 at the hospital. This was a joint effort by the SWF and the Center of Excellence at the hospital. About 30 people attended, including 22 adults and 6 children.

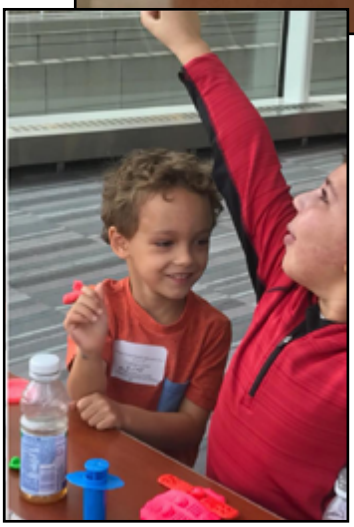
The speakers described treatments and diagnoses and addressed questions and concerns from the families. The day ended with lunch so that the families could connect with other parents and their children.

To allow the adults to focus and listen to the various speakers, the kids were occupied with their own supervised activities, cared for by the hospital's Child Life team.

The expert speakers and their topics were:

- Cynthia Stack, MD neurology
- Hawke Yoon, MD ophthalmology
- Sarah Chamlin, MD dermatology
- Miller Shivers, MD psychology

Kris and Kellie Sadens were the host family for the event. The coordinators at the hospital was Carolyn Kiolbasa, RN, Clinical Nurse for the Vascular Lesions Center and Sarah Chamlin, MD, dermatologist who has been a friend of the SWF for several years.



While the SWF has hosted Education Forums before, this event demonstrates the value of having an institution like Lurie Hospital on board for family education. As our Centers of Excellence expand into the new Clinical Care Network (CCN), it forecasts more joint events that bring both the SWF and the specific hospital's message and mission closer to the families and adults impacted by SWS and port wine birthmarks.

"It's always so wonderful to have the opportunity to connect and learn with new and familiar families in a more intimate setting. We are so excited and grateful for the work the Foundation and doctors across the country are doing to bring accessible, quality care to all SWS families."

Kelllie and Kris Sadens

Pictured to left: Carolyn Kiolbasa, nurse at Lurie's Children's Hospital | Chicago, IL.

Pictured above: These lovely ladies were instrumental in making the Education Forum such a success!

Pictured left: Even the children had a great time at the Education Forum in Chicago, IL.

REPORT: BVMC CONFERENCE

The 2017 Annual BVMC Executive Committee Meeting was held October 31 - November 2 in Washington, D.C. Brian Fisher attended with Webster to discuss existing and future projects for SWF.

The Foundation summarized the outcome of the CCN Conference, Lisa's Research Fellowship Awardee, and plans to hold the next SWFIRN Conference in the Fall of 2018.

Anne Comi, MD and Dough Marchuk, PhD gave a 20 minute presentation the current BVMC project exploring new research in birthmark vascular malformations.

The 2017-18 BVMC Pilot Project was awarded to Jeffrey Loeb, PhD. Dr. Loeb presented his plan for this project which included his IRB and contingency plan. Dr. Loeb also suggested that the search for a new trainee on the project begin immediately.

Pictured above: BVMC Executive Committee

Pictured left: Dr. Loeb and Dr. Marchuk with Webster at the BVMC Conference

REPORT: WHY TISSUE DONATION IS IMPORTANT

We often report on the research on SWS needed to advance the knowledge and treatment of SWS and other rare diseases, and it often sounds distant and unconnected to the everyday life of families and adults. Stories of scientists in their labs and of financial grants awarded to support their work give us all a good feeling, especially when the SWF is part of the funding or participation. "Well, they are working on the problem and soon there will be more answers."

UNIVERSITY OF MARYLAND BRAIN & TISSUE BANK

A Brain and Tissue Repository of the NIH NeuroBioBank

1-800-847-1539

http://www.medschool.umaryland.edu/btbank/family/

But where do ordinary families fit into the picture?

Researchers do not just crunch data, manipulate test tubes and explore specimen slides on their high powered microscopes and devices. They also need actual human contributions in the form of human tissue which can only come from – guess who – humans.

For several years the SWF has been urging families and individuals to participate in our Tissue Donation program. You probably have filed the information in the back of your mental file cabinet.

The thing to remember is that surgery, whether planned or emergent, occupies your thoughts and emotions and leaves little time for thinking about where you filed that information from the SWF.

But somewhere in your medical information file, please consider including the information on SWS tissue donation. We have more details on the SWF website in the "For Patients" section, but the contact to remember is the Maryland Brain and Tissue Bank listed above.

UMBTB has been a leading organization in the initiative to collect and bank vital specimens from individuals living with a rare disease. Their friendly and knowledgeable staff are on hand 24 hours a day in the event of a surgery or sudden death where time is of the essence.

This is not just for brain surgery, but for any surgery that involves human tissue from a patient with SWS or a port wine birthmark. If you are in doubt, the UMBTB can provide the answer.

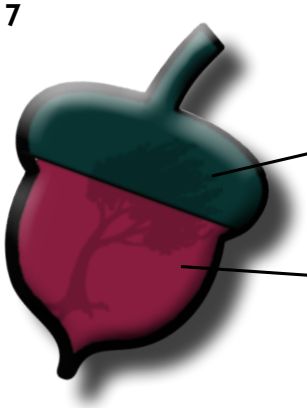
You only have to call and register with the Tissue Bank and they take care of the rest – contacting the hospital, retrieving and tissue, transporting and storing it. But this takes time – and waiting until the patient is in the recovery room may not be helpful.

Families who have donated tissue have told us that this is one way to turn a scary and difficult event into something positive and hopeful.

REPORT: 2017-18 RESEARCH - FIRST QUARTER

SWF RESEARCH FUND
July 1 - September 30, 2017

TOTAL: $133,771.91



Lisa's Research Fellowships
$44,825.25

Research Fund - Donations
$88,946.66

REPORT: CLINICAL CARE NETWORK (CCN) & PATIENT ENGAGEMENT NETWORK (PEN) CONFERENCE

Together with The Sturge Weber Foundation, Members of the Patient Engagement Network (PEN) traveled to New Orleans, LA in October. Keevin Lee, the Clinical Care Network(CCN) and Research Coordinator invited members from all of the Clinical Care Networks to attend as well. In attendance from PEN were 4 parents, 3 adult patients, a sibling and an aunt. From the CCN, there were 17 CCN's represented. The medical team represented Neurology, Ophthalmology, Dermatology, Hematology, Radiology, Pathology, Dentistry and 2 Nurses.

THE STURGE-WEBER FOUNDATION
CLINICAL CARE NETWORK

The goal of the meeting was to create protocols and standards of care in all areas of Sturge Weber Syndrome. The importance in doing this was that we have created a network of doctors that come together and agree on the protocol. We understood that there will be differing opinions but the goal was that we come to an agreement.

As the day began, PEN discussed what we as patients and caregivers need and then discussed each part of the syndrome. As we began each discipline there was an adult patient that told their story in neurology, dermatology, ophthalmology and dentistry. The stories helped both the CCN and PEN to relate to what was needed.

In the afternoon, the breakout sessions began. We had a session for neurology, dermatology and ophthalmology and PEN began to work on Mental Health. The most lively conversation came in Neurology with 9 doctors and a nurse in attendance. The second part of the breakouts talked about Imaging, Integration, and Tissue Banks and Genetics.

The second day was a full morning of updates of what we learned. The CCN's took away that mental health was a larger issue then they knew and we have some work as the PEN on this going forward as this will be the project. The conversations overall were very active and as patients and caregivers it was enlightening to see all these great minds coming together and talking about this very rare syndrome.

We all need our medical teams that we trust to treat our patients individually on a day to day basis. But the biggest take away was that NOT one person can come up with a cure for Sturge Weber Syndrome. It will take a village and after what we saw at this meeting The Village has been created in the Clinical Care Networks and with the guidance of Keevin and Dr. Jeffrey Loeb we will see new papers being written and protocols being put in place. The PEN is very excited to see what will come next.

Top: PEN and CCN members during general session.
Middle: PEN and CCN group photo
Bottom: PEN committee

REPORT: CHIEF SCIENTIFIC OFFICER REPORT

The SWF held its family conference in Cincinnati in July 2017. At that meeting we discussed some topics related to research as we try to get a better understanding of the causes of Sturge-Weber syndrome. Here are five topics.

Publications
Scientists publish papers to share knowledge about what they've learned. Most papers go through "peer review": a research team does experiments, makes some advance in knowledge, writes up a paper, and submits it to a scientific journal. Other scientists serve as anonymous reviewers and usually ask a team to improve their paper in some way (sometimes by doing additional experiments) before the paper can be published. We're eager to see the Sturge-Weber syndrome research efforts grow, and one way to do that is by encouraging scientists to make more discoveries and publish more papers in this area. Here are some recent statistics on the number of papers in PubMed, the main international database for scientific literature.

We can see that there are far more papers published on other diseases such as neurofibromatosis and tuberous sclerosis. (Those conditions also affect more people than does Sturge-Weber syndrome.) We'd like to see more papers published on SWS!

Uveal Melanoma
We all have DNA in our cells that's organized into chromosomes. The DNA is made up of base pairs (in a famous double helix), and it turns out we have about 3 billion bases of DNA in the human genome. And it also turns out that our DNA includes genes—and we each have 20,000 genes that make proteins. We now know that a change in just one base of DNA, in a gene called GNAQ, causes Sturge-Weber syndrome. That one base pair change alters the protein that is produced by GNAQ (that protein is called Gαq and it is activated in SWS).

There's a very surprising connection between Sturge-Weber syndrome and a particular kind of cancer called uveal melanoma. One cause of uveal melanoma is the same mutation in GNAQ—but in a different cell type and at a different time of life. When and where the mutation occurs can change what happens.

We are very interested in understanding what sort of progress the research community studying uveal melanoma is making. They have started clinical trials (although none seem likely to help those with SWS). They have created at least two mouse models (introducing the mutation into mice, creating an animal that has the condition, and then trying to figure out how to treat it). They have found mutations in genes other than GNAQ that cause uveal melanoma—and so we are looking to see if these findings are relevant to people with SWS who we test and who do not have the GNAQ mutation.

Animal Models
Besides mouse models, some groups are creating zebrafish models. On the tree of life, humans, mice and fish are really very closely related—we're all vertebrates, and we share most of our genes in common. So yes there is a GNAQ gene in fish too. It's very likely that scientists will soon have developed a fish model of SWS. One way that's helpful is to screen large numbers of drugs to try to find ones that correct the changes caused by the mutation in GNAQ.

Biobanking
The Sturge-Weber Foundation is working closely with members of the research and clinical communities to create a biobank. That means that when people donate precious tissue—such as a skin punch, or tissue after surgery—we'll have a place ready where we can store the tissue, perhaps grow cells from it, and study it to try to make progress. Putting a biobank in place will be a great way to promote research and to involve more and more scientists in making discoveries.

Genomics
We can look into the DNA of a person to see mutations in GNAQ—or in other genes. Some people wonder if they (or their children with SWS) should get tested by having the GNAQ gene sequenced. Today there's not a need to do that, and it would not impact the clinical care given to a patient.

One company that does a large amount of sequencing is called Macrogen. They've kindly given us a gift to support our work in Sturge-Weber syndrome. Through that gift we're able to do more studies, and we're very grateful!

Do you have questions about the science behind Sturge-Weber syndrome? If you do, email me (cso@sturge-weber.org) or give me a call in the office at (443)923-2686.

Dr. Jonathan Pevsner, Ph.D.
SWF Chief Scientific Officer

MY FOUNDATION | MY FAMILY
FAMILY NEWS

THE WILLIAMS FAMILY

Paul and Nina Williams and their son Jude, who just turned one, along with big brother and big sister, have moved from New Jersey to Belgium. They successfully had Jude's records transferred fromNYC and are scheduled to see Dr. Laurence Boon and her colleagues at University Catholique de Louvain. Dr. Boon, dermatologist, is a long time SWF consultant along with her husband, Mikka Vikkula, a geneticist and researcher.

Jude visited a local garden center with his family.

SWF ANGELS

You may have received an e-mail Holiday card featuring little angels rising with hope in their hearts. This is part of one of our new outreach projects to capture stories of families. You can read the whole collection of stories on the website in the Who We Are or the Participate sections.

The artwork was done by SWF children at the Cincinnati conference in July. It reminds us that our children carry the message of hope to the world, just by being who they are.

If you or your child have a story you would like to include, please contact Susan Finnell at sfinnell@sturge-weber.org.

If you keep HOPE alive,
It will keep YOU alive.

FINISH LINE: 2017 FALMOUTH ROAD RACE RESULTS

As mentioned in the 2017 August/September *Branching Out*, TeamSWF was preparing for its eighth annual FALMOUTH ROAD RACE, led by Pam McIntyre and Jessica Melo, to incredible SWF "Warrior Moms".

TeamSWF did not disappoint! In fact, because of all the runners continued dedication, sponsorships from Liberty Mutual Insurance and Cynosure, they grossed over $146,000 for SWS!

This fundraising event takes months of planning, coordination and dedication to the cause. We are thankful to Pam and Jessica, and their team members for stepping up to the "starting line" every year to continue support and research for SWS. **Congratulations TEAM!**

KIM & MARGOT SLATER

A lovely photo of Kim and her mother, Margot Slater at a recent theater event in West Palm Beach.

MY FOUNDATION | MY FAMILY
PATIENT SPOTLIGHT

Rebecca Stanton

I'm 19 years old and will be starting my sophomore year of college next month. I grew up with a family that was very supportive, and I found that I could rely on my parents and my siblings whenever I was feeling insecure or uncomfortable with who I was. They raised me up and always showed me how to see myself in a positive light. They helped me get through all of the scary things that come with having Sturge-Weber, like laser treatments, surgeries, and sharing with people who don't know anything about Sturge-Weber.

In grade school I let my insecurities get the best of me, not reaching out to other people or joining clubs. Without the comfort of my family being with me at school everyday, I had to make my own decisions and form my own friendships. I was scared and shy, so I decided to focus on building one friendship at a time, getting to know one person and letting them know all about me over time. The one person that I chose to focus on showed me that it is a good thing to open up and share with other people. She became my best friend, and stayed my best friend all the way through high school and to today. She cares not only about me as a person, but also about the issues I face as a Sturge-Weber patient. She always checks in on me after laser treatments for my port wine stain and she is always there to talk about anything.

I spent high school with my one friend, and I believed that was enough. When I got to college, however, I realized that my fear of being judged was irrational and that what really mattered was being happy and connected to lots of other people. I went out of my comfort zone and tried as many clubs as I could, talked to tons of people, and even joined a sorority. I'm excited to go back to school and see all of the new friends I've made, which is something I never looked forward to during my high school or grade school summers.

I learned that having Sturge-Weber can make you feel like you're different from everyone else, but everyone else has something about them that makes them different too. The people I met shared their stories with me, and we were able to learn about each other and enjoy one another's uniqueness. I learned that the fear being different should never limit you from talking to people or doing challenging and exciting things, because more often than not, it will turn out better than you could've imagined. If you reach out to new people with kindness and confidence, you will receive support and love because of who you are.

Brielle Coutu

Brielle is a very very active 2 year old who just loves to climb, jump, swim, dance, listen to music and most of all being outside no matter the weather. She has more energy than all of her cousins put together, but that is what makes Brielle Brielle. She is an inspiration to her family and there is no stopping her.

Brielle started having seizures at age 3 months old. After being put on Keppra and Trileptal to prevent seizures along with aspirin to prevent another stroke like episode and Zantac Brielle stayed seizure free for just under 2 years. This past January she began having seizures again and had at least one episode a month until May when her neurologist Dr. Anna Pinto prescribed her a third anti-seizure medication called Onfi. This third medication has kept Brielle seizure free for two months. In May Brielle had another MRI which showed that the left side of her brain has calcification. Due to this Brielle will be admitted to Boston Children's Hospital for testing in August to help the doctors decide the best course of action to help Brielle stay healthy.

BE A "FRIEND"RAISER

A new word enters our vocabulary and has expansive meaning for our SWF members and supporters. We all know what fundraising is, and many SWF families and supporters have created events and opportunities that have brought financial benefit to the SWF.

But Friendraising is an aspect of raising money for an organization that goes beyond a single event or campaign. It means befriending another person or even an organization with the purpose of helping support the group you are passionate about. Facebook and other social media have taught us how many "friends" are out there.

It does not have to be aggressive or loud. You can start with your own family, friends and neighbors. How many know what Sturge-Weber is? They may know that you or your child have this rare disorder but do they know how it impacts you and your family? Are they reluctant to ask you?

Maybe they respond to the SWF seasonal appeals and then become donors. Do you let them know how you personally appreciate their donation? (Even before they make one?) Do you let them know that they can help spread awareness? Do you ask for more than just money?

Photos from "Friend"raiser event 2017 Falmouth Road Race - August 20, 2017.

Do you let them know how involved you are and invite them to jump on the band wagon with their involvement?

One mom reports that when they moved to a new town, she made a distinct effort to introduce herself and her pre-school son to the neighbors and invite them to playdates at her house. Not only did this minimize the groundless fear of exposing their children to someone with a rare disorder, but when the time came for their family to have a yard sale for the SWF, she already had a group of friends to help promote it.

Some adults with SWS tell us that they grew up not being bashful about their birthmark and it became a signature and an entry to many friendships and associations in later life.

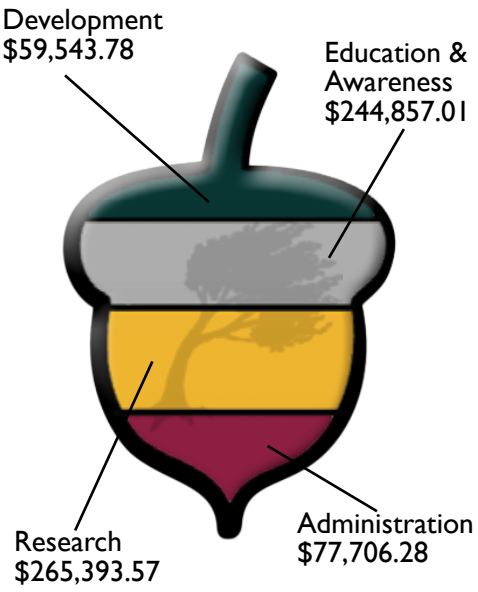
Your friendraising will help not just you and your family, but the whole SWS community of SWF members who will benefit from having a bigger circle of friends.

Other aspects of financial support for the SWF
The Sturge-Weber Foundation also accepts gift-in-kind donations that will help the organization further and fulfill its mission. These include gifts of securities, real estate, or being named beneficiary under trusts, life insurance policies, commercial annuities and retirement plans. (Of course, consult your financial advisor, lawyer, bank, or accountant to find out how exactly to do this).

Our website also has other information and details on ways to be financially generous. Look at the section on Participate – Help Us Help Them. You can donate through the CFC at your workplace, Amazon Smile when you purchase gifts, your car when it has outlived its usefulness to you.

IN A NUTSHELL: 2016-17 YEAR END

GROSS INCOME:	**$711,488.65**
EXPENSES:	**$663,800.57**
NET INCOME:	**$ 47,688.08**



SOCIAL MEDIA: THE GOOD AND THE BAD

Today you hear on the news everyday about Tweeting, Facebook posts and things like "Fake News". The average American can post anything, anywhere. This past summer at the Interntional Family Conference I sat with 3 doctors and 4 other parents and the discussion began about politics. Not about what side we were on in the election, but how do you know what to believe.

Sturge-Weber syndrome is no different. There are many pages on Social Media Sites. On Facebook, there are 103 pages and 90 Groups that have Sturge-Weber syndrome as a part of their page from all over the world. On Twitter, there are 14 Sturge-Weber accounts in some form. If we only look at these two Social Media Sites that is over 200 places to look for Sturge-Weber syndrome.

At first glance, that is truly exciting. Sturge-Weber syndrome affects 1 out of 50,000 children and we have over 200 places worldwide to look at and find advice. If you look deeper, as a caregiver or patient you won't feel alone because you can talk to so many people. You can also ask for advice, look for ideas, it is endless.

Like any project, especially in Sturge-Weber syndrome you need a complete toolbox. While Social Media has many positives it has some drawbacks too. Social Media is comprised of opinions. We also know that every case is different and has many variables. That makes advice on Social Media very challenging. This brings you back to what do you believe?

Social Media if used effectively, can be a great tool. When you seek advice you need to have a team atmosphere, including your doctors and specialists that know your case the best.

The Sturge Weber Foundation, vows that what we post is not "Fake News!" In all seriousness, The Foundation has 30 years experience and knowledge of Sturge-Weber syndrome. We have 25 Clinical Care Networks (CCN) that we work with and have relationships with to learn more.

Let us know what questions and advice you need and we will always do our best to get you the answers you need.

THE STURGE-WEBER FOUNDATION

CLINICAL CARE NETWORK

CA: UC Irvine
Primary Contact: Kristen Kelly MD, Dermatology
CA: UCSP Medical Center-San Francisco
Primary Contact: Ilona Frieden, MD, Dermatology
CA: Rady Children's Hospital - San Diego
Primary Contact: Lawrence Eichenfield, MD, Dermatology
Sheila Friedlander, MD, Dermatology
DE: Nemours/AI duPont Hospital for Children-Wilmington
Primary Contact: Carol Roethke, CRNP-APRN
IL: Ann and Robert H. Lurie Children's Hospital Chicago
Primary Contact: Sarah Chamlin, MD Vascular Clinic Director
IL: U of Illinois at Chicago Medical Center-Chicago
Primary Contact: Jeffrey Loeb, MD, Neurology and Akira Yoshi, MD, Neurology

MA: Boston Children's Hospital-Boston
Primary Contact: Mustafa Sahin, MD, Neurology and Anna Pinto, MD, Neurology
MI: Children's Hospital of Michigan-Detroit
Primary Contact: Csaba Juhasz, MD, Neurology Imaging
MI: U of Michigan Mott Children's Hospital-Ann Arbor
Primary Contact: Jennifer Reeve, MD, Dermatology
MN: Mayo Clinic: Rochester
Primary Contact: Megha Tollefson, MD, Dermatology
NC: UNC Children's Hospital-Chapel Hill
Primary Contact: Craig Burkhart, MD, Dermatology
NJ: Northeast Regional Epilepsy Group-Hackensack
Primary Contact: Eric Segal, MD, Neurology
NY: NYU Medical Center-NYC
Primary Contact: Daniel Miles, MD, Neurology

OH: Cincinnati Children's Hospital-Cincinnati
Primary Contact: Adrienne M. Hammill, MD, Hemangiona and Vascular Malformation
OH: Nationwide Children's Hospital-Columbus
Primary Contact: Warren Lo, MD, Neurology
PR: Centro Medico de Puerto Rico- San Juan
Primary Contact: Rafael Rodriquez Mercado, MD, Endovascular
TX: Cook Children's Medical Center-Fort Worth
Primary Contact: Saleem Malik, MD, Neurology
TX: Dell Children's Medical Center -Austin
Primary Contact: Moise Levy, MD, Dermatology
WA: Seattle Children's Hospital
Primary Contact: Jonathan Perkins, DO, Vascular Clinic Director

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RESPECT
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HOPE

This publication is brought to you by the generous contribution of the Allergan Foundation.