The Sturge-Weber Foundation
12345 Jones Road, Suite 125
Houston, TX 77070

believe
you. us. progress.

A PUBLICATION OF THE STURGE-WEBER FOUNDATION

Branching Out

SEPTEMBER | OCTOBER 2019

973.895.4445 | www.sturge-weber.org | swf@sturge-weber.org

The Sturge-Weber Foundation
MAGAZINE

CONTACT INFORMATION
12345 Jones Road, Suite 125
Houston, TX 77070
973-895-4445
swf@sturge-weber.org
www.sturge-weber.org

DESIGN/EDITOR
Susan Finnell

EDITORIAL CONTRIBUTIONS
Karen L. Ball, SWF CEO
Anne Howard, SWF Writer
Susan Finnell, Marketing|Home Office

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

September | October 2019

On the Cover: Riley Gutierrez

IN THIS ISSUE . . .

Port Wine Birthmarks (PWB) on the skin are developmental abnormalities in blood vessel formation (capillary malformations) that are more extensive and darker than the pink capillary birthmarks often seen at the nape of a baby's neck.

Sturge-Weber syndrome (SWS) is a rare congenital condition usually consisting of a facial port wine birthmark, glaucoma, and seizures, (although not all of these symptoms may be exhibited).

SWF POLICY STATEMENT:

In implementing the purpose of The Sturge-Weber Foundation to improve the quality of life for individuals with SWS and their families, the Foundation will act as a clearinghouse of information, provide emotional support and facilitate research on PWB and SWS.

The Foundation will seek information regarding management and treatment techniques and suggestions concerning education and emotional support and will facilitate the dissemination of appropriate information.

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

THE STURGE-WEBER FOUNDATION CLINICAL CARE NETWORK

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

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

Illinois:
- **Ann and Robert H. Lurie Children's Hospital**
  Primary Contact: Sarah Chamlin, MD-Vascular Clinic Director
- **University of Illinois at Chicago Medical Center**
- **Primary Contact:** Jeffrey Loeb, MD, Neurology
- Akira Yoshi, MD, Neurology

Massachuessets:
- **Boston Children's Hospital**
- **Primary Contact:** Mustafa Sahin, MD, Neurology
- Anna Pinto, MD, Neurology

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

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

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

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

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

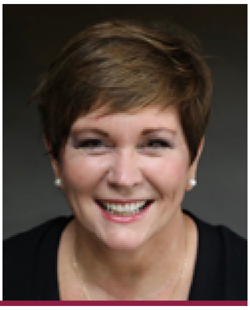
Ohio:
- **Cincinnati Children's Hospital-**
  Primary Contact: Adrienne M. Hammill, MD, Hemangiona and Vascular Malformation
- **Nationwide Children's Hospital-Columbus**
  Primary Contact: Warren Lo, MD, Neurology

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

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

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

For updates on list of CCNs, go to sturge-weber.org to "For Patients" and click on Clinical Care Network.



THEY SAID

Karen L. Ball
Founder and CEO

They Said: You've got your hands full with a sick baby you can't start an organization (1987 $0 --2019 $813,000)

They Said: You're working and raising a handicapped child you shouldn't add a fundraiser to your plate too. (Pennsylvania proud for 25 years)

They Said: Stick with softball it's what you know but a Gala was born (Houston Proud)

They Said: You're too rare for corporate pharma to support you (Reunion of Champions Lead On)

They Said: You started with a cookbook and moved on to a Road race just don't tackle too much. ($1,000,000 total raised in 10 years)

They Said: You'll never find the gene that causes SWS because it's not inherited. (GNAQ gene mutation - 2013)

They Said: This new generation of millennials needs a medal (Indiana, Illinois, Texas, California, Florida, Massachusetts, New Jersey, to name a few- Warriors Deliver)

They Said: There's a lot of competition out there for researchers to find a cure (Zebrafish makes a splash - 2019)

WE SAID: Believe…and we WILL make an impact!

We've all heard the phrase used often enough. It's as if the person making the pronouncement just by the shear fact they are stating it makes whatever they are commenting on a truth or a done deal! Really…come on. I've always been one who, when I hear that phrase, immediately revert to "well watch this!" unless of course it's from an admired source who is your own personal cheerleader and "They Said: You can do whatever you set your mind to!"

I'm happy to report I've been fortunate to be surrounded by others of the same mindset! Which in turn means you are fortunate too because without them the SWF would not have made all the strides we have since 1987.

The 2018-19 annual report will soon be out and please take a moment to peruse it. Lots of exciting events and programs have exploded and it will highlight what's ahead. Thank YOU for doing your part in your corner of the world to make an impact!

EDUCATION DAY CONFERENCE

October 5, 2019 | 8:00 AM - 2:00 PM
Ann & Robert H. Lurie Children's Hospital of Chicago
225 East Chicago Avenue | Chicago, IL 60611

If you are in the area and would like to attend, please register by contacting Carolyn Kiolbasa, RN | 312-227-8521
email: vlc@luriechildrens.org.

There will be activities for the children so that parents can focus on the important information provided. The conference will end with lunch together and the opportunity to visit and connect with other patients and families.

Conference Speakers

Dr. Sarah Chamlin, Dermatology
Dr. Hawke Yoon, Ophthalmology
Dr. Miller Shivers, Psychology
Dr. Cynthia Stack, Neurology
Dr. Jeffery Loeb, Neurology, SWF Chief Clinical Strategist

Ann & Robert H. Lurie
Children's Hospital of Chicago

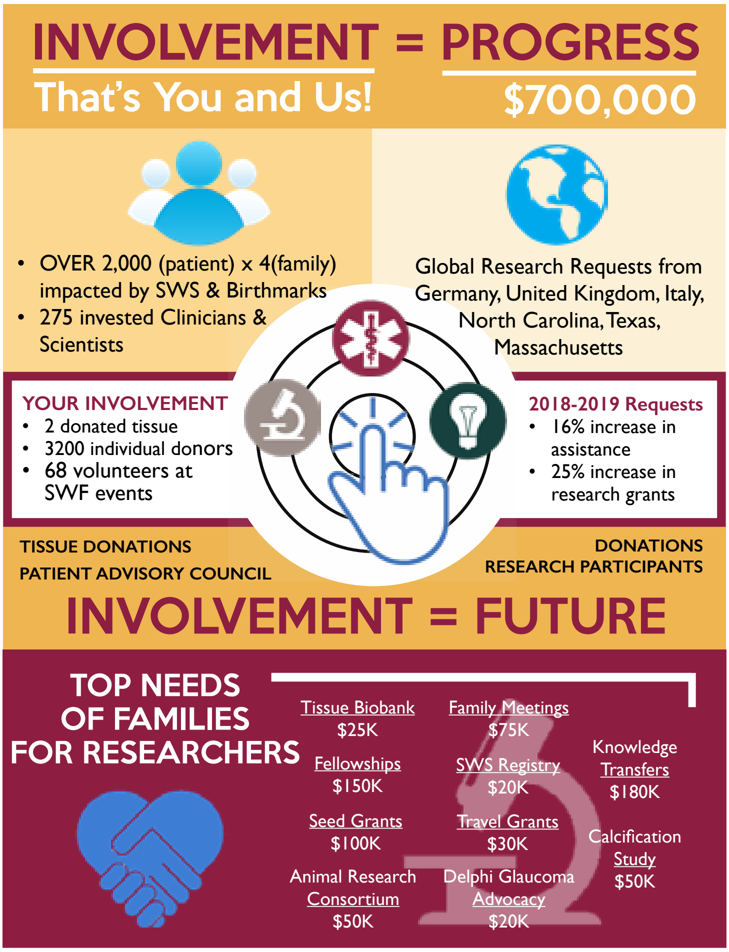
RECAP: 2019-2020 BOARD MEETING INVOLVEMENT=PROGRESS!

In July, the Board of Directors met at the 2019 International Family Conference. They welcomed new member, Witney Arch, to her first meeting and voted in Curt Stanton as Chairman. Estelle Benson, founder of Gullian Barre Syndrome Association, shared some good ideas for governance and volunteer expansion with the SWF Board.

L-R: Witney Arch, Jeffrey Needham, Estelle Benson, Karen Ball and Curt Stanton (not pictured Woody Crouch, Kremena Linguiti and Kris Sadens)

The Board then reviewed the past fiscal year's progress to strategically plan and set goals for 2019-2020. The Foundation has many exciting opportunities this year that will push research forward and facilitate international collaboration, more grant opportunities for SWS and PWB fellowships, tissue biobank, family meetings and more.

Check out our goals to the right!



INVOLVEMENT = PROGRESS

That's You and Us! **$700,000**

- OVER 2,000 (patient) x 4(family) impacted by SWS & Birthmarks
- 275 invested Clinicians & Scientists

Global Research Requests from Germany, United Kingdom, Italy, North Carolina, Texas, Massachusetts

YOUR INVOLVEMENT
- 2 donated tissue
- 3200 individual donors
- 68 volunteers at SWF events

2018-2019 Requests
- 16% increase in assistance
- 25% increase in research grants

TISSUE DONATIONS
PATIENT ADVISORY COUNCIL

DONATIONS
RESEARCH PARTICIPANTS

INVOLVEMENT = FUTURE

TOP NEEDS OF FAMILIES FOR RESEARCHERS

Tissue Biobank	$25K
Family Meetings	$75K
Fellowships	$150K
SWS Registry	$20K
Knowledge Transfers	$180K
Seed Grants	$100K
Travel Grants	$30K
Calcification Study	$50K
Animal Research Consortium	$50K
Delphi Glaucoma Advocacy	$20K

FAMILY, FRIENDS & FUN!

RECAP FROM THE 2019 SWF INTERNATIONAL FAMILY CONFERENCE

Thursday morning, July 18, 2019, SWF staff and volunteers dispersed like an army on a mission preparing for the 2019 Family Conference. This conference was different in so many ways from years past - 2 different locations; 2 conference tracks - pediactric and adult patients; a dinner cruise; movie night and SWF International Research Network Conference facilitated at the same time.

Thursday, registrants began to arrive to check in to the hotel, stop by the conference registration desk and prepare for the 2019 Route to a Cure Walk. Delaware weather was not cooperating that day - it was 98 degrees and raining! While those of us from Houston, TX are quite familiar with heat and humidity, plus the complexity of the syndrome and the inclement weather, we decided to cancel the walk, but were able to tour the beautiful duPont mansion.

Following the tour, an old fashioned ice cream social was held for families, speakers and doctors to visit with one another.

Friday, tour buses were ready and waiting to carry everyone over to the Nemours/Alfred I. duPont Hospital for Children, for a full day of valuable information on the latest research and available treatments. In the afternoon, the conference tracks were divided into pediatrics and adults. This provided more specific information and Q&A time for each age group. Children spent the day with Corporate Kids playing games, doing crafts and building new friendships.

Following a full day, families were given the opportunity to spend free-time or join the Foundation for a movie night which included all your favorite movie snacks, art and crafts, and selfie station!

Saturday consisted of wrapping up the conference, then off to a time of respite for all caregivers! Women got to have their nails and make-up done, while the men were able to play golf or just hang out at the golf club. This time of respite was provided by a generous donor who wanted to make sure that caregivers were provided an opportunity to rest and relax a little while the children were in a safe childcare environment! We thank you for your generous and thoughtful gift!

Finally, we were off to the dinner cruise upon the Spirit of Philadelphia along the Delaware River. It was a night of dinner, dancing and celebrating our journey together. Truly, another wonderful conference experience. This event is like no other. It is an opportunity to learn, reconnect, and celebrate each other, together.

THANK YOU FOR BELIEVING IN US!

PROFESSIONALLY SPEAKING

RECAP FROM THE 2019 SWF INTERNATIONAL FAMILY CONFERENCE & 2019 SWF INTERNATIONAL RESEARCH NETWORK CONFERENCE

It was a rare opportunity to facilitate the 2019 SWF Family Conference and 2019 SWF International Research Network Conference at the same time. Thanks to our Clinical Care Network Center, Nemours/ Alfred I. duPont Hospital for Children, patients, families and professionals were able to interact like never before, affording opportunities to see daily life for patients and caregivers, while seeing the vast new potential for research progression.

SWF is so thankful to the speakers and professional volunteers that played a major role in making these conferences possible. We especially are grateful to Dr. Stephen Falchek, Director of Neurology, Carol Greene-Roethke, CRNP-APRN, Supervisor for Neurology; Sean Fennington, Marketing Specialist and Jennifer Hultberg, Neuroscience Administrative Coordinator *(pictured at beginning of article)* who worked diligently along-side Julia Terrell, Conference Director and the SWF staff.

Here is a small snapshot of who and what was discussed at the conferences:

SWF International Family Conference

- Tissue Repository - Dr. Thuy Phung
- Dentistry and You - Dr. Sarat Thikkussey
- CBD Discussion - Dr. Stephen Falchek; Dr. Anna Pinto; Dr. Jeffrey Loeb
- Glaucoma - Dr. Jonathan Salvin
- Surgery: Is It For You? - Dr. Harry Chugani
- Transitions - Dr. Amy Renwick
- Endocrinology - Dr. Dan Doyle
- IEP Planning - Debbie Esposito
- Transitions to Adulthood - Josephine Suarez

SWF International Research Network Conference

- Vascular Anomalies: Disease Models and Targeted Therapies - Dr. Elisa Boscolo
- Novel ex vivo Model of Vascular Anomalies - Dr. Thuy Phung
- Modeling Rare Vascular Diseases in the Zebrafish - Dr. Nathan Lawson
- Phenotypic Outcomes of Mosaic, Prenatal Signaling Pathway Activation Depend on Cellular Competence - Dr. Heather Etchevers
- Imaging Brain Abnormalities and Plasticity in SWS Understand Brain Abnormalities in Children with SWS - Dr. Csaba Juhasz

Pictured on this page - Dr. Anna Pinto, Dr. Jeffrey Loeb and Dr. Thuy Phung

Survey Says . . .

Q: What was the best part of the Conference for you and your family?

A: Meeting new friends and reconnecting with old ones. Having new experiences and going new places.

A; Learning more about SWS.

A: Really enjoyed the last minute addition of the Nemours mansion tour since the Walk was cancelled due to inclement weather!

THE STURGE-WEBER FOUNDATION
10
ROOTS TO A CURE®

YOU. ARE. AMAZING!

2019 Falmouth Road Race | 10 Years of Faithful Support

by Susan Finnell

The year was 2009. Two SWS mothers, Pam McIntyre and Jessica Melo, of Massachucetts set out to hold their first fundraiser. They created a cookbook that generated $9,800 in donations. The following year, a tiny team of 7 was created which became 78 strong this year! "That first year in Falmouth, I would have been hard pressed to find 7 people who knew what Sturge-Weber was, never mind articulate its effects. Now, each and every runner, and so many generous sponsors and donors, are true ambassadors for this rare disease", says Pam McIntyre.

> **"Great things are done by a series of small things that are brought together."**
>
> **Vincent Van Gogh**

Each year as the team grew, the donations grew and awareness of SWS grew. This year alone Team SWF set a goal of $260,000, exceeding that total by $27,000. Combining all 10 years of faithful support, over $1 million dollars has been raised by this grassroots team. Indeed, there is truth in great things are done by a series of small things, brought together!

RECAP: 2019 TEAM SWF FALMOUTH ROAD RACE

Top: 2019 Team SWF running team, 79 strong! Left: Jessica Melo and Pam McIntyre, fearless leaders for 10 years. Below: Dr. Anna Pinto crossing the finish line!

The Foundation would not be able to continue assisting new and existing patients and families and research without faithful donors. 2,316 individual donors supported the cause within a 6 month period. 72% of the donors have not donated before, 38% are faithful returning donors. Many of the returning donors have been supportive all 10 years - Liberty Mutual, Amica, CREA, Orix, Cynosure, Grantham, Mayo, Van Otterloo & Co., Woburn Police Department, and so many, many others.

I had the honor of being at the race this year and shaking the hands of many of the participants. It was so heartwarming to hear them tell me what an honor it was for THEM to run the race for Team SWF. The extended family and sense of community this event has created can only be described as incredible. Together anything is possible. Step by step, momentum is created and progress is achieved!

Check out more photos! →

believe
you. us. progress.

PREGNANCY AND CHILDBIRTH SURVEY

The SWF has never done a formal survey that asks our members to share information about their pregnancy and childbirth experiences. There is now an opportunity to participate in the knowledge base that all can benefit from.

We thank Stephanie Tikkanen, who is the aunt of Jayden Findlater and a professor at the School of Communication Studies at Ohio University for developing our questions into a useful survey. Also SWF members Rebecca Szorcsik and Michelle Daoust, for their help in asking the right questions. Dr. Anna Sarafina of University of Illinois in Chicago provided information on seizure medications during pregnancy.

If you are a woman who has SWS or PWB and you have had a successful pregnancy and childbirth, no matter how recent or long ago it was, please take this survey. It will provide important information and may give some young adults the confidence that they can have a successful life experience. If you are the parent of a young woman who has SWS or PWB, please share this notice with her.

The survey was sent via our August E-news and can be seen on the electronic version of Branching Out which includes the link https://ohio.qualtrics.com/jfe/form/SV_9mBz46FbUwCjJR3. Or you can contact Julia Terrell at jterrell@sturge-weber.org and ask her to send you the link.

Anne Fischer, PH.D., pictured here with Jen, is Jen's favorite professor.

GROWING GOLDEN: Caring Comes Full Circle

Often adult children experience a turn-around of roles and become the chief caregivers of their parents as life dictates changes, challenges and issues. But it is uncommon that an adult with SWS experiences this role reversal. As adults with SWS become independent and assume typical responsibilities, they also may develop a special empathy for the mother or father who saw them through the medical and emotional turmoil of their growing up with SWS.

Jennifer Berg and her mom Roberta live in Florida and have been with the SWS since 1998 They have come to several conferences (Orlando, Denver) and have always been eager to participate and be part of the SWF family. Jen has just achieved her bachelor's degree after a persistent journey when she put her goals on hold to care for her mom's needs.

JENNIFER'S STORY

My name is Jennifer Berg and I have Sturge-Weber Syndrome, but that is not who I am.

I have had a total of 71 surgeries, 2 brain surgeries, 6 eye surgeries, wearing a prosthetic eye, mouth surgery and many face surgeries. My mom has been through all of it with me. She has always supported and taken great care of

me. That has made us close.
In high school I received my degree as a certified CNA, then after high school I obtained my AA degree.
In 2014 I started going to the University of South Florida. But shortly thereafter, my mom got very ill. She was diagnosed with cancer, had surgery, was cleared, but then 5 months later it came back, stage 4, after that it came back two more times.

During all this she was in bad shape and could not do anything, even walking was difficult without my help. So, I quit school and became her sole caregiver (role reversal). I felt that with all the medical events I had been through and watching her take care of me, it was only natural that I take care of her. It was an easy decision.

Once she got better (she is now cancer free) I returned to school. I finally obtained my Bachelor of Science and Mathematics in Psychology from the University of South Florida.

I am aware that parents of children with medical problems have a life-long job, and most of them are great at it. As a parent, you know your child better than anyone. Don't always assume that doctors know everything and if you feel something is wrong or your child or teen feels something is just not right, LISTEN. Sometimes you might just have to change doctors to get the right answers and treatments.

Also, always let your kids try things that interest them, in school or out, they just might surprise you.

I want to put all my experiences to good use working with children, especially those with disabilities, mental or physical. I know I can relate to their problems and will be able to communicate and understand what they are going through.

ROBERTA'S STORY

Being the mother of someone born with Sturge-Weber has not been easy. Jennifer is an adult now and just graduated from USF with a bachelor's degree in mathematics & science in psychology. I am so proud of her.

All through school, her teachers put her down and kept telling her she would never really amount to anything. But she has always been determined and strong willed. Plus, I have always told her that she can be and do whatever she chooses.

She arrived 2 weeks early and only weighed 2lbs, 13oz. Her first surgery was at 3 months of age for glaucoma. Her second was at 7 months for the same thing. Also, back then they really didn't know anything about

Sturge-Weber, at least down here they didn't. I even had one doctor that wanted to treat her ask me how to spell it. So much for that doctor.

When she was 23 months old, she had her first seizure and it lasted for 1 hour and 45 minutes. The doctors even came out to me and said she was going to die. Little did they know what a fighter she was and still is.

I have been a single mom to Jennifer since she was three months old. It has never been easy, but I would not change the experiences and challenges we have faced over the years. As of this date, she has had 71 surgeries, including 2 brain surgeries. Just watching her grow and overcoming all the obstacles has been painful and yet rewarding.

When she was in high school, she obtained her CNA certificate, was in the marching band (the drum line), karate, police and sheriff's cadets, she was on her bowling team for 11years. Also, starting in elementary school, she was counseling other students. Teachers actually called her out of class to come to their room and help with someone. She always came through and the student was calmer and more focused after having been with her.

With all the medical events she has been through, she really has learned so much. A few years ago, I got extremely ill with cancer that kept coming back, Jennifer dropped out of school and became my fulltime caregiver and advocate. She gave up everything to take care of me and get me back to being a real person, even when the doctors had given up on me.

I also found out that she is quite a good cook. I am so blessed to have her in my life, and I know that it was mostly because of her and all the prayers. It took over two years (and no complaining from Jen) but we got through it. I guess you could say we came full circle, I took care of her through all her surgeries, hospital stays and all the ups and downs of growing up and in turn, when I needed her, she was right there taking care of me, no questions asked. I feel very blessed to have such a remarkable young lady in my life.

BETTER TOGETHER

Catching Up With the SWF Family

The recent Growing Golden article in Branching Out by Ann Nehrbauer about her family’s experience has acted like a pebble in a pond - sending of ripples.

One bit of updated information is from Linda Cohen, mom of Marco and his brother Roberto. She shares photos of Marco at his school prom, and one of Roberto, who will be going to the University of Chicago and taking his cello with him.

Linda reports that she recently was at Willowbrook which has been re-purposed for educational seminars and workshops. Her words say it best.

“I almost started crying after I read Ann Nehrbauer’s story of her son Stephen in the last issue. I visited Willowbrook for 5 days of training in Self Direction for Marco recently and it has been re-purposed and reclaimed, but watching the videos of the past was hard. But then again, you realize how resilient the human spirit can be. We also heard from people who lived through all that - like Stephen - and now live independently and are happy. Thanks to all those at Willowbrook who advocated for their rights, our kids now have other choices like Self Direction and the HCBS waiver.”

SWF ON TISSUE DONATIONS

An always important and needed activity all families can participate in is the SWF Tissue Donation program. Scientists need human tissue when they delve into finding out the how and why of SWS and PWB, and, as important as animal models and computer projectsions are, there is no substitute for human bodily tissue.

It could be cells from a recent lip reduction, or dental procedure. Or, on the extremely serious end, tissue from a hemispherectomy. The SWF works with the Maryland Brain and Tissue Bank to help secure this valuable resource.

Their office procures the tissue from the hospital and keeps it in a repository. The donor's family do not have to do anything except notify the Bank of an upcoing (or emergent) surgery. But before that, the family can register with the Bank so that their information will be already on file. No need to fill out forms when you have enough stress because of a coming surgery.

You can get forms needed from the SWF and send them to the Maryland Brain Bank. Contact Julia Terrell at jterrell@sturge-weber.org to get them and for additional information.

UNIVERSITY OF MARYLAND BRAIN & TISSUE BANK

A Brain and Tissue Repository of the NIH NeuroBioBank