A MAGAZINE FROM THE TUBEROUS SCLEROSIS ALLIANCE

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Tuberous Sclerosis Alliance

MESSAGE FROM THE LEADERSHIP

Kari Luther Rosbeck
President & CEO

Beth Lewin Dean
Chair, TS Alliance
Board of Directors

Throughout 2019, the Tuberous Sclerosis Alliance (TS Alliance) will celebrate our sapphire, 45th anniversary. This momentous milestone, chronicled so beautifully in this issue's cover story, is only possible due to the tenacity of untold numbers of tuberous sclerosis complex (TSC) community members, physicians, researchers, staff and board members, partner organizations and other supporters who – from 1974 to 2019 – dedicated their lives to ensuring the TS Alliance stays laser-focused on our mission to find a cure for TSC while improving the lives of those affected.

As we kick off 2019, we first want to welcome several new Board of Directors members, including Matt Bolger; Steven Goldstein; Heather Lens; Darcy Krueger, MD, PhD; Mustafa Sahin, MD, PhD; Sean Shillinger; and, for our Endowment Fund, Marion Adams. We look forward to their input and expertise. We'd also like to salute outgoing Board members Rebecca Anhang Price, PhD; Michael Caggiano; Martina Bebin, MD, MPA; and Laura Lubbers, PhD; and from our Endowment Fund, Rita DiDomenico. Thanks to each of you for your heartfelt guidance and commitment during your years of service on the Board.

The TS Alliance recently updated our Strategic Plan, which will guide our efforts for the next five years. Before doing so, we sought community input through an online constituent survey. You can read more about the entire process on page 16. In a related effort, the TS Alliance also recently released an aggressive five-year research business plan to support an effort to raise an additional $56 million to speed up TSC research. You'll find more details about it on page 24.

We know how critical research is to the TSC community, and many of you help further these efforts by participating in clinical trials. The TS Alliance also gets many questions about current studies being conducted, so we've included a partial list on page 26. If you'd like to find out even more about clinical trials, please visit www.tsalliance.org/clinicaltrials where you can view a video, download a brochure and view the entire list of current TSC clinical trials.

Of course, the TS Alliance is always pleased to share success stories in *Perspective*, and this issue is no exception. On page 15, TSC mom Lisa Hall provides a 10-year update on her daughter's successful epilepsy surgery. Then, on page 18, another TSC mom, Julie Comeaux, explains her and her son's sometimes excruciating fight with their local school system, ultimately demonstrating the importance of being an advocate for your children.

Special events play important roles in our fundraising and community-building efforts. In 2019, we've updated our Step Forward to Cure TSC® walk program, so we hope you'll find a way to participate in an event near you. In addition, we look forward to the 18th Annual Comedy for a Cure® in Hollywood on April 7, the 3rd Annual Sound Bites in Minneapolis on May 10 and the 45th Anniversary Gala on October 3 in New York City. We are grateful for each event's volunteer chairs and committee members who work so hard behind the scenes.

In 1974, four mothers who shared the common bond of tuberous sclerosis complex came together to provide fellowship, generate awareness, pursue more knowledge and offer hope to each other. They founded the TS Alliance on the core belief that community is a sustaining strength in the face of difficult challenges. Forty-five years later, their spirit and intention still influence everything we do. We hope you will each join us as we celebrate them and the many, many successes we've seen over the years. But we know much work remains, and the TS Alliance will strive – always – to find better treatments, offer improved support programs and ultimately, one day, build the blue brick road to a future where no one has to endure the devastating effects of tuberous sclerosis complex.

PERSPECTIVE
WINTER 2019 · VOLUME 146

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If you have opinions, questions or articles for *Perspective*, we would like to hear from you. Please contact the managing editor to obtain a submissions form and guidelines.

Perspective is intended to provide basic information about tuberous sclerosis complex. It is not intended to, nor does it, constitute medical or other advice. The Tuberous Sclerosis Alliance (TS Alliance) does not promote or recommend any treatment, therapy, institution or health care plan. Readers are warned not to take any action without first consulting a physician. Commentary expressed herein reflects the personal opinions of the author and does not necessarily reflect the official views of the TS Alliance. Information contained in the TS Alliance database is confidential and not provided nor sold to third parties.

Perspective is published by the National Tuberous Sclerosis Association, Inc. d/b/a Tuberous Sclerosis Alliance, a 501(c)(3), charitable organization.

Congress Enacts $6 Million for Tuberous Sclerosis Complex Research Program in Fiscal Year 2019

On September 26, the House cleared the final House-Senate conference report to the fiscal year 2019 Defense/Labor-Health and Human Services-Education Appropriations Act, which includes an additional $6 million for the Tuberous Sclerosis Complex Research Program (TSCRP) at the Department of Defense (DoD). The Senate had previously cleared the conference report on September 18, and the final package was signed into law on September 28.

Thanks to the advocacy efforts of the TSC community, Congress has consistently appropriated $6 million annually for the TSCRP. Since fiscal year 2002, Congress has appropriated an aggregate amount of $83 million for the TSCRP!

Through this same appropriations package, Congress also approved an additional $2 billion increase for the National Institutes of Health (NIH), another important funding source for TSC research. In the past three years, Congress has increased the NIH's budget by a total amount of $7 billion, a remarkable accomplishment in this tight fiscal environment.

116th Congress Sworn In Amid Partial Government Shutdown

On January 3, the House and Senate members of the 116th Congress were sworn into office, commencing a new session of Congress. However, the important business of finishing all twelve appropriations bills was never completed by Congress, and, as a result, many departments and agencies have been shut down since December 22. One of the first items on the agenda is approving a continuing resolution to re-open the parts of the federal government that have been affected by the shutdown.

It is important to note that many agencies are NOT affected by the partial government shutdown, including the Departments of Defense and Health and Human Services. As a result, the TSCRP and NIH continue to operate under normal procedures, and research grants on tuberous sclerosis complex are not impacted by the shutdown.

March on Capitol Hill Scheduled for March 5-7

The Tuberous Sclerosis Alliance will once again sponsor our annual "March on Capitol Hill" from March 5-7, when advocates from across the country will be coming to Washington, DC to personally advocate for continued Federal funding for the TSCRP. For more information, please contact Katie Smith at (301) 562-9890 or ksmith@tsalliance.org.

The TS Alliance is also pleased to announce a new champion for our "Dear Colleague" letter. With the retirement of Representative Ileana Ros-Lehtinen at the end of the 115th Congress, Representative Markwayne Mullin (R-OK) has agreed to serve as the lead Republican member on the House Dear Colleague letter in the 116th Congress. We deeply appreciate Representative Mullin's support over the years, and his commitment to serve in this new leadership role.

IN MEMORIAM: SUE (MARGARET SUSAN) POVEY, 1942 – 2019

by David J. Kwiatkowski, MD, PhD, Brigham and Women's Hospital, Boston, MA

In 1991, I attended my first international meeting on tuberous sclerosis complex in Nottingham, England. It was a very special event for me as I had the opportunity to meet all the current and many future leaders in TSC clinical care and research, including Manny Gomez, Vicky Whittemore and Sergiusz Jóźwiak to name a few. I also met Professor Sue Povey at that meeting, which was particularly special.

Sue was a lifelong geneticist dating back to her college days at the University of Cambridge. She joined the staff of the MRC Human Biochemical Genetics Unit, University College London, where she rose to become Deputy Director, and Haldane (a legendary geneticist) Professor of Human Genetics. Sue made very important contributions to TSC research for many years. She was a key investigator on the first report of genetic linkage to chromosome 9, beginning the study of the genetics of TSC; she contributed in a major way to the discovery of the TSC1 gene; and she worked on the TSC diagnostic criteria and management guidelines published in 2012. Arguably most importantly, she set up and supervised the LOVD TSC mutation database for the past 15 years, maintained by Rosemary Ekong. This resource is used on a regular basis by numerous TSC researchers, clinical geneticists and patients around the world.

But I will remember Sue best for her wonderful positive attitude and cheerful willingness to share samples and data freely towards a common goal, something made very clear in our collaboration that led to the identification of TSC1 in 1997. I will always remember her as a wonderful role model in science and as a friend.

Tuberous Sclerosis Alliance
STEP FORWARD®
TO CURE
TUBEROUS SCLEROSIS
COMPLEX

In 2019, **Step Forward to Cure TSC®** gets a fresh new look as we continue to salute our Super Heroes and TSC Champions across the country! Over the past 17 years, the TSC community, volunteers, corporate partners and advocates have helped raise nearly $15 million through Step Forward to Cure TSC walks, helping fuel research progress and breakthroughs. For example, TS Alliance-sponsored research helped identify the two genes that cause TSC, developed a genetic test, initiated the first clinical trials in TSC, and supported the first studies aimed at preventing seizures from ever developing in TSC.

Again, we want to thank our national 2018 Step Forward to Cure® TSC sponsors, our local walk chairs, walk committees, donors and supporters for making our 15th year a year to remember for the TS Alliance:

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Additional Step Forward to Cure TSC National Walk on the Mall Sponsor

Mallinckrodt
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Fall 2018 Step Forward to Cure TSC Results

TS Alliance of the Carolinas

Raised: $12,371
Date: September 8, 2018
Location: Mint Hill Veterans Memorial Park, Mint Hill, NC
Walk Chair: Heather Morris

TS Alliance of Indiana

Raised: $11,301
Date: September 15, 2018
Location: Billericay Parkl, Fishers, IN
Walk Chair: Pat Schmutte

TS Alliance of Oklahoma

Raised: $13,824
Date: September 15, 2018
Location: LaFortune Park, Tulsa, OK
Walk Chair: Heather Lens

TS Alliance of New York and New Jersey

Raised: $30,232
Date: September 22, 2018
Location: Wantagh Park, Wantagh, NY
Walk Co-Chairs: Carol Ann White and Lori DiStefano

TS Alliance of New England

Raised: $32,222
Date: September 22, 2018
Location: Auburndale Park, Auburndale, MA
Walk Chair: Stacie Verrill

TS Alliance of Western Pennsylvania

Raised:$8,757
Date: September 29, 2018
Location: North Park Harmar Pavilion, Allison, PA
Walk Co-Chairs: Christine Koch and Debbie Koch

TS Alliance of Intermountain West

Raised: $18,110
Date: September 29, 2018
Location: Murray City Park, Murray, UT
Walk Chair: Rob Thurston

TS Alliance of Florida (Miami)

Raised: $9,123
Date: September 29, 2018
Location: Florida International University, MMC Campus, Miami, FL
Walk Co-Chairs: Vanessa Vazquez and Danny Jackson

TS Alliance of New York and New Jersey

Raised: $15,586
Date: September 30, 2018
Location: Oak Ridge Park, Clark, NJ
Walk Chair: Tresha Bisang

TS Alliance of St. Louis and Southern Illinois

Raised: $7,040
Date: October 13, 2018
Location: Jackson City Park, Jackson, MO
Walk Chair: Donna Bullard

TS Alliance of Nevada (Las Vegas)

Raised: $2,061
Date: October 13, 2018
Location: Craig Ranch Regional Park, North Las Vegas, NV
Walk Chair: Kathleen Kingston

TS Alliance of Alaska

Raised: $1,738
Date: October 13, 2018
Location: Virtual
Walk Chair: Rebecca Racenet

TS Alliance of Wisconsin

Raised: $10,170
Date: October 6, 2018
Location: Regner Park, West Bend, WI
Walk Co-Chairs: Scott Brown and Geri Greenberg

TS Alliance of Northern California
Raised: $29,807
Date: October 27, 2018
Location: Marina Park, Seagull Picnic Area, San Leandro, CA
Walk Chair: Brittany Britt

TS Alliance of Kentucky/ West Virgina
Raised: $4,406
Date: October 20, 2018
Location: Des Pres Park, Louisville, KY
Walk Chair: Emilie Berryman
Planning Committee: Pam Howard and Krista Merriman

TS Alliance of Nevada (Sparks)
Raised: $3,738
Date: October 28, 2017
Location: Del Cobre Park, Sparks, NV
Walk Chair: Tara Zimmerman

TS Alliance of Tennessee
Raised: $ 17,124
Date: October 27, 2018
Location: Edwin Warner Park, Nashville, TN
Walk Contact: Gail Saunders

TS Alliance of Florida (Tampa)
Raised: $44,219
Date: October 27, 2018
Location: Lowry Park, Tampa
Walk Co-Chairs: Amy Hightower and Jamie Jodoin

TS Alliance of Ohio
Raised: $6,326
Date: October 28, 2018
Location: Cleveland Metroparks The Chalet
Walk Chair: Lori Quirino:lquirino@roadrunner.com

Step Forward to Cure TSC®
2019 Spring Walk Schedule

Location	Date	Where	Walk Chair(s) / Regional Program Manager
Arizona	April 6, 2019	Scottsdale, AZ	Debora Moritz & Terri Butler / Julie Scroggins
Texas (Houston/Austin)	April 27, 2019	Houston, TX	Rachael Jackson / Gail Saunders
Pacific Northwest (Seattle)	April 28, 2019	Renton.WA	Gloria Triebenbach & Adrienne Wasserman / Julie Scroggins
Connecticut	May 4, 2019	Manchester, CT	Jennifer Waldron / Shelly Meitzler
DC Metro (National Walk on the Mall)	May 5, 2019	Washington, DC	Lauren Shillinger / Gail Saunders
Texas (Dallas)	May 11, 2019	Carrollton, TX	Sylvia Sparby / Gail Saunders
Alabama	May 18, 2019	Russellville, AL	Jessica White / Gail Saunders
Georgia	May 18, 2019	Dacula, GA	Lauren Perry & Jeff Treimer / Gail Saunders
Illinois	May 18, 2019	Galesburg, IL	Karen Johnson-Wenger / Julie Scroggins
Pennsylvania (East)	June 1, 2019 (pending)	West Chester, PA	Jack Lyons / Shelly Meitzler
Colorado	June 8, 2019 (pending)	Northglenn, CO	Julie Scroggins
California (South)	June 8,2019	Long Beach, CA	Shannon & Rob Grandia / Julie Scroggins
Michigan	June 8,2019	Lansing, MI	Treasa Bolger Dunlap / Shelly Meitzler
Ohio (Cincinnati)	June 9, 2019	Evendale, OH	Nathalie Mortine / Shelly Meitzler
Illinois (Chicago)	June 22, 2019 (pending)	Mundelein, IL	Jacqueline Wolak / Julie Scroggins

Keys to the Cure TSC Champions

The TS Alliance welcomes and encourages all levels of community fundraising through the **Keys to the Cure** program. These events and initiatives aren't just about raising money; they are also about spreading awareness in your local community and providing everyone the opportunity to actively participate in the mission of TS Alliance.

To view current and upcoming initiatives or events, please visit www.tsalliance.org/keystothecure.

4th Annual Dance for Austin

Raised: $6,209
Date: September 8, 2018
Location: American Legion Post 40, Glen Burnie, MD
Contact: Jaime Edwards

13th Annual Friends and Family BBQ

Raised: $8,280
Date: September 8, 2018
Location: Bill and Nancy Youmans' Residence, Lakewood, CO
Contacts: Bill and Nancy Youmans

KEYS TO THE CURE

12th Annual Heather Joy Memorial Golf Tournament for TSC Champions

Raised: $6,185
Date: September 8, 2018
Location: Salem, WI
Contacts: Loren and Christy Buntrock

Kick TSC to the Curb Kickball Tournament

Raised: $7,311
Date: September 8, 2018
Location: Franklin Park, Prarie Village, KS
Contact: Laurisa Ballew

2nd Annual Team Lyons Pride Roaring to Cure TSC

Raised: $8,000
Date: September 14, 2018
Location: Barnaby's Aston, Media, PA
Contact: Jack Lyons

Corn Maze for a Cause

Raised: $1,100
Date: September 29, 2018
Event Location: Brookfield Pumpkin, Thurmont, MD
Contact: Lauren Shillinger

Corn Maze for a Cause.

15th Annual Tournament for TSC Champions

Raised: $2,468
Date: October 6, 2018
Location: River Glen Country Club, Fishers, IN
Contact: David Cowlin

Rollin' for a Cure

Raised: $ 2,026
Date: November 3, 2018
Location: Bilou Roller Skating Rink, Fulton, NY
Contact: Bridgett Claxton-Langstaff

Facebook Donations / Birthday Fundraisers

$127,664 raised through December 31, 2018!

Thank you for remembering the TS Alliance on Facebook to mark your birthday.

Tuberous Sclerosis Alliance
TEAM TSC
TRAIN • SWEAT • CONQUER

43rd Annual Marine Corps Marathon, Washington DC Results

The Tuberous Sclerosis Alliance, supported by Team TSC, was proud to be an official charity partner at the 2018 Marine Corps Marathon with 19 registered runners, who **raised $50,000** for our organization! The team was led and motivated by Team TSC Ambassadors Jill Woodworth and Ryan Novotny. Special thanks to our 2018 Team TSC members: Kerilyn Amedio, Ali Blanchette, Dave Carpenter, Rob Carpenter, Kate Carter, Rob Cronin, Seamus Donoghue, Dana Imperatore, Olivia Imperatore, Ryan

Novotny, Kaila Parrott, George Petty, Kay Rawlings, Casey Ryan, Taylor Ryan, Beth Wilson, Micheal Wilson, Jill Woodworth and Eugene Zebrowski, Jr.

2018 TCS New York City Marathon Results

For the second year, the TS Alliance had a dedicated group of 11 individuals running on our behalf in the TCS New York City Marathon. Special thanks to our 2018 Team TSC members: Wendy Anderson, Rob Bendini, Joseph Cervantez, Susan Cohen, Brianah Doeden, Bridget Faria, Justin Largent, Allen Newbauer, Stephen Presley, Ashley Roth and Kiran Thomas. These runners have been actively fundraising and training for the largest marathons in the country. The team has **raised more than $69,000** to advance the mission of the TS Alliance. We were grateful to have Celia Mastbaum and Liz Buchsbaum host what has become an annual Rooftop Celebration the day before the marathon, where a small group of local TSC family members came together to eat, make signs and celebrate Team TSC.

If you're interested in joining Team TSC in 2019, visit **www.tsalliance.org/team-tsc** today**.**

TSC CHAMPIONS SPOTLIGHT:

2018-2019 Junior Leaders

Kierstin and Dalton Dublinske.

Dalton Dublinskie

My name is Dalton Dublinske, I'm 17 years old. I have two sisters: Chesney (8 years old) and Kierstin (13 years old). My sister Kierstin is diagnosed with TSC. TSC, of course, has been a huge part of my life, and so as soon as I heard about the Junior Leader program, I knew this would be something I would love to do. I'm very excited to start helping out everyone in the community, and it's such a pleasure to be accepted for this position.

Brianna Johnson

Brianna Johnson.

My name is Brianna Johnson and I am 21 years old. I was diagnosed with TSC at the age of 3. I live in Scottsdale, AZ and am in my senior year of college at Grand Canyon University. In my free time, I enjoy taking photos, crafting, and watching sports.

Emilie Peters

My name is Emilie Peters. I am 21 years old and from Owasso, OK. I was first diagnosed with TSC at 8 months of age. Shortly after my diagnosis I began having infantile spasms. Over the years I have faced many challenges and had many different types of seizures. Maintaining seizure control has been a constant struggle. I have now been seizure free for 14 weeks on my current medications. Even though I have always strived to lead a normal life, it has required help from my family, teachers, and many others. My parents and older sister have always been a great support for me. I graduated high school in 2016, and currently attend Tulsa Community College where I am pursing a degree in Human Services. I hope to be able to take my experiences and use those to assist other families in dealing with children with disabilities.

Emilie Peters.

Justin Leslie

Justin Leslie.

Hello, my name is Justin Leslie. I was diagnosed with TSC at the age of 10 with a very mild version of the disorder and am driven by my "luck" to spread awareness about the disease because not many people know about TSC! I am a pharmaceutical science major at the University of Rhode Island. Having TSC is the driving force as to why I want to work for a pharmaceutical company when I am older, and I hope to give back to the people who are less fortunate than I am. I applied to be a Junior Leader because I truly believe spreading awareness about TSC and fundraising for a cure are the best possible things I can do right now to help. The Tuberous Sclerosis Alliance helped me by giving me mentors and keeping me in the

SAVE THE DATES: UPCOMING MAJOR EVENTS

18th Annual Comedy for a Cure®

Sunday, April 7, 2019

Co-Hosts: Alex Skuby and Jim O'Heir
Emcee: Melissa Peterman

Comedy for a Cure® is set to return for its 18th straight year on Sunday, April 7, 2019 at Academy LA on Hollywood Boulevard. Since its inception, Comedy for a Cure as raised more than $4.75 million! The event begins with a one-hour reception and silent auction at 5 pm, a brief program on TS Alliance progress and advancements, and our much-anticipated live auction with auctioneer extraordinaire, Damon Casatico, followed by an all-star comedy lineup featuring John Henson. This year, the **Szylagi Family** will be presented the Courage in Leadership Award for the compassionate dedication they have provided the organization since their 23-year-old daughter, Emily, was a young child. The event's long-time comedian supporter, **Wendy Liebman**, will be honored with a TSC Champion award as will **Dr. Joyce Wu** of UCLA for her ongoing support for the advancement of treatments and therapies for TSC. For more information, please visit www.comedyforacure.org.


The Szylagi Family.


Wendy Liebman.


Dr. Joyce Wu.

Third Annual Minnesota Sound Bites

Friday, May 10, 2019

Honorary Co-Chairs: Chris Hawkey and Kat Perkins
Event Chair: Kari Ihle
Emcee: Melissa Peterman


The Spotts Family.

The TS Alliance's **Third Annual Sound Bites, An Evening of Food, Wine, and Music,** will return to The Metropolitan Ballroom & Clubroom in Golden Valley, MN, on Friday, May 10, 2019. A change of seasons and a weekend setting promises to take this fabulous event to the next level after a wildly successful event in 2017. The event showcases the best area restaurants and wineries who donate their items and service to provide guests with a unique experience. **The Spotts Family** will be awarded the Courage in Leadership Award for their dedication and leadership, while **Carol Hoskin, RN**, and **Upsher-Smith** will both receive TSC Champion Awards for their generosity in supporting the advancement of treatments and therapies for TSC. A special shout out goes to the locally grown talent, Chris Hawkey, Kat Perkins and Melissa Peterman, who will put together another really awesome show. Their support, drive and personal commitment to the success of Sound Bites have helped to make this a successful area event. For more information, please visit www.tsalliance.org/SoundBitesMN.


Carol Hoskin, RN.

CELEBRATING 45 YEARS OF CHANGING THE FUTURE IN THE FIGHT AGAINST TSC

Cover Story

Inspiration | Innovation | Hope
45 YEARS OF CHANGING THE FUTURE
1974
2019
Tuberous Sclerosis Alliance

2019 is a major milestone for the Tuberous Sclerosis Alliance as it marks our organization's sapphire anniversary. To highlight the occasion, we've chosen the theme, "45 Years of Changing the Future – Inspiration | Innovation | Hope."

In 1974 with nowhere else to turn to help their children with TSC, four mothers from Southern California – Debbie Castruita, Adrianne Cohen, Susan (Diaz) McBrine and Verna Morris – joined together to lay the foundation to form the TS Alliance. Their vision to provide fellowship, generate awareness, pursue more knowledge and offer hope to anyone affected by the disease still guides the organization today. With their examples, the TS Alliance has now transformed into an internationally recognized organization dedicated to finding a cure for tuberous sclerosis complex while improving the lives of those affected.

Here, two of our founders and other community members walk us through each decade, starting with the 1970s, to demonstrate this organization's ongoing commitment to inspire, innovate and create hope for anyone touched by tuberous sclerosis complex.

The 1970s

Adrianne Cohen

I am so happy to help commemorate the 45th year of our mission with the families and individuals of the Tuberous Sclerosis Alliance from my viewpoint of the early 1970s.

I have always had great difficulties sharing my personal story that led me to be one of the organization's founding moms. Talking about the early times makes me think about what I didn't know, didn't do, or should have done or could have done at that time.

As one of the founding moms I am proud to share my thoughts about the grassroots beginnings of the National Tuberous Sclerosis Association, now known as the TS Alliance. The four founding moms never hesitated to support one another, and we began our mission to find other families with TSC, to offer information and to begin to direct parents to medical care.

Ronda and Adrianne.

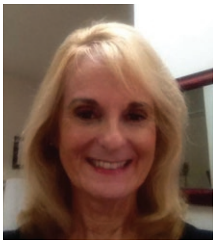
I personally realized significant advances would not come soon enough to help my child, Ronda, and others. Therefore, in the beginning we knew we had to plan and persevere with very little information and technology, or science and research that was not yet available to us. In spite of these challenges, we all felt the urgency of our children's needs; we were hopeful.

In the beginning families were isolated and alone. One of the first immediate goals we had was to reach out and contact families. One turning point for the organization was meeting Dr. Manny Gomez who did TSC research at the Mayo Clinic. He published the first definitive major study of tuberous sclerosis complex in 1979. Our registry evolved from this meeting, which provided documentation for further research. Grants and funding research followed; medical conferences and information were shared. But most of all, his work gave credibility to our efforts and validated the need for our organization.

Our friendships gave us a shared sense of strength; we developed a deep empathy for other families; we gave each other hope for the future. This 45th anniversary is momentous indeed. It reminds us to inspire each other and acknowledge the day-to-day suffering and urgency that is still present.

What helps me sleep at night is knowing that Tuberous Sclerosis Alliance is still working each and every day to find a cure. This organization continues to provide hope!

Susan McBrine



More than 45 years ago in 1972, I was a first-time mom experiencing every mom's worst nightmare in an emergency room: my 9-month-old baby's diagnosis of an incurable rare disease, tuberous sclerosis complex, about which doctors knew very little. They predicted the very worst-case scenario and very a limited life span for my daughter, Stacia.

I refused to accept this prognosis and immediately began searching libraries for information. In the 70s, there was no internet or email, and research on TSC had never been done. That meant our options to communicate and/or learn more consisted of letters, phone calls, books and periodicals. The MRI was not even available until 1973, and genetic testing and diagnosis were limited, not to mention treatment! Moreover, it often took many years to obtain a proper TSC diagnosis back then.


Stacia and Susan.

As an educator, I was aware of existing support groups like The ARC. I also soon discovered *Exceptional Parent* magazine. In my desperation to find other cases of TSC, I held out hope that, with some numbers, we could join together to increase awareness, to promote research, to get doctors to diagnose and treat TSC faster and better and, most of all, to know we would not be *alone* facing a TSC diagnosis. So I wrote a letter to *Exceptional Parent* looking for other parents. This letter led me to three other California moms: Adrianne Cohen, Verna Morris and Debbie Castruita. Their TSC children were 9, 19 and 4 at the time, while mine was almost 3 by then. All had debilitating cases.

The four of us shared perspectives, dreams and even our anger at our children's fate. We combined our contacts and our skills to make our vision a reality and we worked hard! We met, talked, planned and slowly, but surely, an organization began to take root and, over decades, spread its wings to become the Tuberous Sclerosis Alliance, which is now revered internationally.

We wrote letters, made phone calls, met with doctors and researchers, wrote and published a newsletter, and created a survey to begin a database. Adrianne secured a grant, so we got a lawyer and filed to become an official nonprofit: the National Tuberous Sclerosis Association (NTSA). The NTSA grew slowly but surely by helping other moms and dads one by one and group by group; by reaching doctors and educating professionals; by holding the very first family conference; and by organizing a medical advisory board.

Today, the TS Alliance's 45th Anniversary's vision of inspiration, hope and innovation certainly enhances our original dream to support families facing the same TSC diagnosis back then. As one of the founding moms, I am so very thrilled to know that vision will be ongoing for years beyond my lifetime! The success of the TS Alliance is a testimony to all the families and individuals who continue to refuse to give up and who also continue to advocate for their child or family member living with the agonies of TSC. Knowing parents and adults can now access instant support – via the TS Alliance website, social media or a phone call as well as now having access to the critically important TSC Clinics – is truly a culmination of our beginnings in the 70s!

We moms who founded this organization did so out of sheer desperation and frustration. But now, thanks to the TS Alliance, no one needs to feel desperate or frustrated due to a lack of support or hope!

The remaining founders are now grandparents, and our TSC adult children all eventually lost their lives to tuberous sclerosis complex. It means so much for us to now know so many people with TSC are living normal life spans and healthier lives with a better quality of life. It makes my heart happy and gives my daughter's life and death more meaning to know every parent and adult can get support and that TSC research continues.

The TS Alliance will certainly endure for another 45 years and beyond until a cure is found. Thank you to the many, many others in the decades following our beginnings who worked so hard to continue to grow the organization into what it is today!

The 1980s

Linda Ham

Our daughter Margo was diagnosed with tuberous sclerosis complex at 10 months of age in 1981. We were referred by our pediatrician to a pediatric neurologist who confirmed she was having infantile spasms due to TSC. We had never heard of it before, nor had anyone else we knew. This doctor suggested we do not do any further research as it was bound to upset us. (At the time, Dr. Manny Gomez's book on TSC had not been published and there was no parent booklet either.) I went directly to my local hospital library and found very little information. One of the sited articles was actually dated 1897! It stated the average lifespan for someone with TSC was estimated at four to seven years.

Hope soon arrived with the referral to the National Tuberous Sclerosis Association (NTSA) from my local pediatrician. Within two weeks of Margo's diagnosis I was standing in the home of Jeff and Gloria Benham. Jeff was the newly elected president of the NTSA Board of Directors, and his home was only an hour from mine. I will never forget looking into the eyes of parents who understood my desperation.

My husband and I knew we had to stay connected to this organization if we were ever going to find out the latest treatment options. I volunteered to serve on the Board and to respond to all correspondence. The organization's mail began being forwarded to my home in September. Jeff and I started to contact all Chicago-area hospitals, physicians and others who might be serving our children. We held numerous parent meetings, trying to determine if there were enough cases locally to create a volunteer base in our area. With each meeting more and more families attended, contacts were made and the NTSA headquarters eventually moved from California to Illinois.


Margo and Linda.

Did you know Ann Landers dedicated a column to TSC? In January 1983 this article appeared, and I was awakened with the first calls from the East Coast. The phone rang non-stop for days. We needed recruits to take turns answering the phone. There were so many calls it became clear all we could do was take names, addresses and phone numbers with the promise we would respond as soon as possible. At the end of three weeks, NTSA had almost quadrupled the number of known diagnosed cases and gained thousands of new contacts including grandparents, other family members and friends. We now had a national corps of potential volunteers and a significant funding base. We also responded to 1,500 people whose children had seizures for no known diagnosis and were able to rule out TSC for them.

My mentor, Dr. Peter Huttenlocher, was a true inspiration. He agreed to start the first monthly clinic dedicated to TSC. Through our parent meetings it was clear there were no standard treatment protocols. There was little consistency in treatment, the advice given or the anticonvulsant regimes. Dr. Huttenlocher agreed to start a database seeking consensus and best treatment options. (In the early days this often included a long list of "what not to do.") He was also willing to accept calls from other physicians if their patient had been seen at least once at our local clinic.

One example that comes to mind is a Wisconsin family we invited to the clinic. They had recently returned from London after seeking a new treatment for infantile spasms. Upon examination of this baby and reviewing EEGs and photographs, Dr. Huttenlocher agreed to treat up to six babies for infantile spasms with vigabatrin. Six cases were identified in less than two months, all of whom responded to the treatment. Dr. Huttenlocher was the only doctor in the nation to my knowledge who was willing to follow these patients, even though their prescriptions were filled in Canada. His clinical interest, expertise and caring demeanor were a true inspiration to everyone.

All the accomplishments I was privileged to witness and now the many, many more that I read about today must be credited to the parents, families and friends of TSC. They have universally been unwilling to simply accept this diagnosis, so they carry their stories to anyone who will listen, while they make great personal sacrifices to lobby, volunteer, fundraise and plead with physicians, geneticists, researchers to take up our banner. Without their passion, there is no story!

The 1990s

Scott Johnson

As the 90s commenced, my wife, Emily, and I were approximately six years into our unwelcomed adventure with tuberous sclerosis complex. Our daughter, Annie, had been diagnosed at 15 months following an onset of seizures. Until then, she had been a typical infant/toddler. Her development had been well within "normal" expectations.

For the years following that diagnosis, she would have two to three seizures each week, her intellectual development would slow, and she would exhibit obsessive and anxiety prone behaviors, along with the "fog" of multiple anti-convulsive meds. And, she would become more and more separated from her chronological peers.

For most of the 90s there was no worldwide web, no Google to search, no TS Alliance website; there was simply not much useful or accessible information about the disease. What you could find, primarily in medical journals/libraries, generally described only the most severely affected of what we now know to be the wide TSC spectrum. We felt alone and unprepared for what might lie ahead.


Annie and Scott.

Through sheer serendipity via a mutual friend, Emily discovered that there were other parents in our town (Santa Rosa, CA) with children with the same disease. Remarkably, they were somehow connected to a national organization associated with it, the NTSA. That was our introduction to the Morris Family (Verna being one of the founding four mothers) and the Mlynarczyks (Laura being an early President of the organization). Finding others dealing with many of the same issues and uncertainties we were now facing was life-saving. These two couples, along with several others to whom we were soon locally introduced, were inspiring examples of how to deal with what life throws at us, turning those challenges into activism.

Within months, Emily took on the responsibility of NTSA "State Rep," a precursor to today's Community Alliance chairs. In this role, she provided support to newly diagnosed families, directing them to resources as well as organizing local medical conferences, family picnics, and fundraisers. If there has been one thing good about having a child with TSC, it has been the deep and lasting friendships we developed with other parents, volunteers, physicians and staff members.

Two important events occurred late in the decade. One was the first National Family Conference in Washington, DC, in 1998. It was a chance to connect with so many other families as well as to hear the very latest in TSC standards of care, emerging therapies, new research, etc. from the talented clinicians and researchers dedicated to finding a cure while managing/mitigating TSC's various manifestations. We learned more in those three days than we had in the prior ten years. We left better informed, further inspired and rightfully hopeful.

The other important event was the discovery of the TSC genes and eventually a genetic test to determine whether one carried the TSC markers. Since Annie has an older sister, Megan, this would later become critical to Megan's decision to have children of her own—or not. Happily, we now have three healthy grandchildren. But this information, regardless of whether the tests are positive or negative, has been hugely beneficial and consequential to thousands of families such as ours.

As the 1990s ended, my Mother, Shonnie Johnson, was concluding her term as a Board member and my Dad, Ken Johnson, ended the same as an Endowment Fund Board member, though both would remain active, in one way or another, forever. Emily had passed her coordinator baton to another mother with a more recently diagnosed child. It was finally time for me to retire as spectator and accept a Board nomination, inspired by all those folks I had been exposed to for the last decade and more, to join a remarkable group of folks on the 2000 TS Alliance Board of Directors, where I would have the privilege of serving initially as a Board member and later its Chair, which was one of the great honors of my life.

Epilogue: Following 20 years of two to three seizures per week, Annie was determined to be an excellent candidate for seizure surgery. Developments in imaging technology provided the surgeon the precise location of where the seizures emanated. She had the surgery in 2003 and has been seizure free since!

The 2000s

Debora Moritz

My son, Griffin, was born in late 1997 when treatment for tuberous sclerosis complex was reactive, and the course of action was primarily "wait and see." Clinicians typically provided grim prognoses, and parents like me were left to fend for themselves in the unknown. The National Tuberous Sclerosis Association (NTSA) was a singular lifeline of support and information.

The identification of the genes TSC1 and TSC2 in the late 1990s set the stage for scientific advancements in the new century in the understanding of tuberous sclerosis complex. Knowing the gene products (the proteins tuberin and hamartin) and something about their role in regulating the critical cellular pathway, mTOR, finally provided targets for treatment. We were moving out of the world of "wait and see" into a brave new world of action – potentially prevention and perhaps a cure.

The NTSA soon changed its name to the Tuberous Sclerosis Alliance. What's innovative or inspirational or hopeful about a name change? It was a philosophical shift, a change in operations to reflect the importance of engaging and forming alliances with the people affected by TSC; the researchers, scientists and clinicians; and other organizations. Alliances are more about mutual interest and benefit and taking action. The TS Alliance recognized this and began to focus the power of the people.

The Community Alliances were also born at this time. Grassroots advocates across the nation now had a vehicle for action. Signature events like the Comedy for a Cure® and the Step Forward to Cure TSC® walks were created by volunteers from the "bottom up," providing crucial funding for TS Alliance operations and research. The first-ever Federal funding specifically designated for TSC research in the Congressionally Directed Medical Research Program (CDMRP) came about because of one grandfather, Will Cooper, making the initial ask and gathering the support of individuals affected by TSC. Individuals were empowered to take action in fundraising, advocacy in education, research directions, raising awareness and providing support and hope.

The first decade of the new century brought much hope for improved clinical care and better lives for the individuals with TSC. Talented researchers and clinicians became even more passionate in their work as the TS Alliance brought them together to define standardized diagnostic criteria and clinical guidelines for care and to build a research plan for the future. All of this was done with the support and coordination of the TS Alliance and the engagement and involvement of the TSC community members, patients and families.

The most inspirational part of that first decade was watching the growth in grassroots advocacy. It was an awakening of the TSC community, a recognition of the importance of their "voice" and a realization of the strength they could draw from one another. Just as clinical care transitioned to action and prevention, so too did the individuals living with TSC move to action. They would not "wait and see" if research would be funded. They would tenaciously advocate for it. They would not "wait and see" if a clinical trial would work. They would step up and participate. They would not "wait" to be asked. They would step forward and do whatever it takes.

The people who will improve lives while finding a cure for TSC are the informed, empowered individuals living with TSC. And the TS Alliance is always there with information and support.

The 2010s

David Fitzmaurice

Happy New Year to everyone! My wife, Jeannine, and I are honored to Chair the TS Alliance's 45th Anniversary Gala in New York City this October. The theme for this event is "45 Years of Changing the Future: Inspiration, Innovation and Hope." As 2019 begins, we reflect on the progress of our own journey with our son, John, who was diagnosed at birth with tuberous sclerosis complex. The progress we have witnessed first-hand with this disease over the last 7.5 years is incredible. To our family, the TS Alliance has been our foundation. They advocate upon our behalf, keep our family informed and educated, and inspire us to participate in the advocacy of the disease as time allows.

The Fitzmaurice family.

Inspiration: TS Alliance President & CEO Kari Luther Rosbeck has helped build an incredible team at the TS Alliance. The culture within the organization is one where everyone selflessly works day and night to help move the mission of the TS Alliance forward. This team of individuals, in coordination with the Board of Directors and the hundreds of community volunteers, should inspire us all to join in the mission. The TS Alliance is a small grassroots organization that has accomplished many amazing milestones over the years, but the future is what excites us the most.

Innovation: We always tell people we know John is in a better place today because of the dedication and effort of so many people who have served before us. The TS Alliance has been at the forefront of patient advocacy and research for some time and is a model organization in the rare disease space. With a better understanding of the science behind TSC, the TS Alliance continues to push research into unchartered waters. Our son, John, has benefited from two recent breakthrough drugs approved over the last ten years. Vigabatrin (Sabril®), which was FDA approved to treat infantile spasms, has controlled John's seizures since he was 5 months old. Rapamycin (sirolimus) cream has also helped to control the angiofibroma growths on his face. Many other people have benefited from another FDA-approved treatment: Afinitor®, a drug developed to treat SEGAs, angiomyolipomas and as adjuctive therapy for partial onset seizures associated with TSC. To build upon these incredible developments over time, the TS Alliance has recently debuted a new, comprehensive five-year Research Business Plan with the goal of raising $56 million by 2023 to speed up research in TSC. This is an incredibly aggressive initiative we hope will change the course of the disease even faster.

Hope: At times, TSC can rear its ugly head. It affects all of us in different ways, and our family tries to live one day at a time. We try not to sweat the small stuff we may have before the diagnosis. We celebrate what to some may seem insignificant, but to John the little victories mean the world. The research funding the TS Alliance provides and advocates for gives our family and others hope. In what has been discovered and developed and for what is to come – the TS Alliance brings hope for a better tomorrow to many individuals and families.

Please join us in celebrating the many milestones the TS Alliance has accomplished over the years. These achievements have improved John's life and the lives of so many others affected as well. There may not be a cure today, but with continued research, collaboration amongst other peer groups led by the TS Alliance and community advocacy, we are confident there will be a day when we find a cure.

Peter B. Crino, MD, PhD

I saw my first tuberous sclerosis complex (TSC) patient in 1992 as a neurology resident at the University of Pennsylvania. At this time, it was just "tuberous sclerosis" or "TS," there were no TSC genes yet identified, no formalized or consensus diagnostic criteria published, and no mTOR link, and amazingly, Facebook, iPhones, and Wikipedia were still in the future, a decade away. I went to review the neuropathology of my patient following epilepsy surgery, and I became fascinated by the effects of TSC on brain structure (cortical tubers), which opened an entire career of scientific investigation for me. But as I started seeing more TSC patients during my fellowship and then as an attending physician, what captivated me most were the courageous, poignant, often heart-rending stories related to me by patients and their families, each detailing a journey from the fateful day of diagnosis to sitting in my office that day.

For so many individuals, the journey was dark and frightening, starting with wrong assessments, for example, that infantile spasms were just "weird baby things," to often heartless opinions – my favorite being, "Well, I guess you can cancel the plans for Harvard" (true story) – to simply uninformed conclusions that all individuals with TSC will be "retarded" (as was often the parlance in the 1980s and 1990s). Countless tales of failed medications, unsuccessful (or worse) unnecessary brain surgeries, kidney surgeries, lung surgeries, diet therapies both real and fad, hospital admissions, difficulties at school, difficulties at home, challenges holding jobs, making friends, paying bills, starting families. So many journeys and so many obstacles, for so many individuals. And yet, these brave souls traveled on.

Like many new to the field, my first real contact in TSC was Vicky Whittemore, PhD, who I will be forever grateful to for encouraging my interest in TSC, helping to get my first grant funded, and connecting me to the then National Tuberous Sclerosis Association (NTSA). Vicky was a "North Star" for me and for so many patients with TSC by guiding us to see another side to the journey, one where I could right so many of the wrongs encountered along the way by simply giving patients realistic facts about outcomes, names of complicated disorders like lymphangioleiomyomatosis in TSC, and strategies to cope with this complicated disease. I realized by becoming an expert in TSC, I could make the unknown a little less scary, the darkness a little brighter, and by providing a "North Star" to patients on this journey, I could do a lot of healing even if I wasn't curing TSC. I could provide hope and inspiration in places where there hadn't been much.

With the establishment of the Tuberous Sclerosis Alliance (TS Alliance) in 2000, on the heals of dual TSC gene discovery in 1994 and 1997, momentum began to build with much more broad awareness about TSC as a multisystem complex, a clear link with a druggable cell signaling cascade, and new diagnostic imaging technologies to view disease progression. The TS Alliance was absolutely mission-critical to the expansion of public health awareness about TSC, providing an infrastructure for designated TSC clinics around the United States, each helping to serve as "North Stars" for patients and families with updated information, protocols for diagnosis and surveillance, and plans for follow up and care. In addition, the organization helped to provide research funding for TSC and to lobby our legislators for enhanced funding across multiple platforms such as the NIH and Department of Defense. The TS Alliance brought researchers and clinicians together in the same room so fewer and fewer stones were left unturned.

The arrival of Kari Rosbeck as the organization's President & CEO in 2007 provided new energy, excitement, acumen, and savvy to our organization, bolstering staff with scientific, communications, and lobby expertise, for the first time positioning us at the forefront of national discussions about epilepsy, autism, and cancer. The darkness was lifted for many and we were out in front as a paradigm disorder providing one of the first examples of precision medicine in autism, epilepsy, and tissue overgrowth syndromes.

The TS Alliance's efforts provided hope and inspiration, as well as expertise, brainpower and funds, to fuel innovation. From these amazing efforts, a drug target was identified and mTOR inhibitors provided the first class of drugs with therapeutic potential in TSC. To be sure, mTOR inhibitors are not a cure for TSC, far from it, but they offer hope and promise to a journey that is often fraught with obstacles, disappointments, and despair for many. The TS Alliance now has sister organizations around the globe, such in Canada, Portugal, and China to name just a few, who have begun to provide hope and inspiration much like the TS Alliance. Amazingly, as I have visited these sites, the TSC journeys traveled by patients and families everywhere often share common themes of fear, sadness, and confusion. The TS Alliance's world-wide outreach is now working to erase these challenges and to provide support for some of the most tenacious and courageous individuals I have ever had the privilege to meet.

The journey that is TSC is not just traveled though by patients and families. Every person who cares for TSC patients, researchers trying to solve basic disease mechanisms in TSC, advocates who fight for rights, support, and funds for TSC families, and lay individuals who come to understand what this diagnosis means are irreversibly touched and set on their own TSC journey. It is impossible not to be drawn into this journey when you see the twists and turns the pathway takes. The path is brighter now, with clear directions, a destination, places to rest, and happily places to stop and enjoy a view of health. There is still a journey for every TSC patient, but our community has worked so hard to make it better for all, and the TS Alliance is the brightest "North Star" in the sky.

TS Alliance to Host 45th Anniversary Gala in New York City

On **Thursday, October 3, 2019**, the Tuberous Sclerosis Alliance will mark our sapphire anniversary, celebrating *45 Years of Changing the Future through Inspiration, Innovation and Hope*, during an intimate **45th Anniversary Gala** at the Edison Ballroom in New York City. We hope you'll plan now to join us as we honor and highlight historical milestones and individuals who helped pave a "blue brick road" in our search for better treatments, improved support programs and one day, a cure for tuberous sclerosis complex.

The 45th Anniversary Gala starts at 6 pm with cocktails and hors d'oeuvres followed by tributes to our honorees, a brief TS Alliance presentation, and an exciting live auction. Throughout the evening, we'll also provide glimpses into our past through special video presentations and music. Long-time TS Alliance supporter Jim O'Heir (*Parks & Recreation*) is our Honorary Chair, and we are pleased Melissa Peterman (*Reba*) will join us as the evening's emcee.

During the 45th Anniversary Gala, we'll pay special tribute to the following who have made a tremendous impact on the TSC community:

The Engles Family, ***Courage in Leadership Award***, for their generosity in advancing TSC collaborative research, scientific breakthroughs, and clinical care while encouraging trailblazing philanthropy.

In 2016, Gregg and Molly Engles created the Engles Collaborative Research Fund, following a pledge of $1.5 million. This transformational gift provided crucial resources to accelerate TS Alliance collaborative research, including the Preclinical Research Consortium and the Biosample Repository. Since their gift, 22 new compounds have been tested, six experiments on model characterization or biomarkers have been run and 756 biosamples have been collected. The Engles were also founding members of the TS Alliance's President's Council, encouraging other TSC families to generously invest in TS Alliance research to dramatically advance scientific progress. The Engles Family Foundation additionally supports TSC research at Brigham and Women's Hospital looking at causes and treatments for TSC-derived LAM (impacting primarily women with TSC) and access to quality care through gifts to the Herscot Center for Tuberous Sclerosis Complex at Massachusetts General Hospital and Children's Colorado. Their extraordinary commitment, inspired by their daughter Lucy, will no doubt improve the lives of everyone affected by tuberous sclerosis complex.

Greenwich Biosciences, ***TSC Champion Award***, for leading-edge integration of the patient perspective into the fabric of their company and pioneering therapy development.

Greenwich Biosciences is at the forefront of cannabinoid science, leveraging nearly two decades of pioneering investment and research by their parent company GW Pharmaceuticals plc. Their focus is to unlock the potential of cannabinoid prescription medicines to address serious conditions that currently have limited or no treatment options. Their commitment to scientific rigor and exacting pharmaceutical manufacturing standards enables them to bring forward plant-derived cannabinoid prescription medicines for patients. Greenwich Biosciences is the first and only company to pursue and receive FDA approval for a plant-derived cannabinoid therapy, Epidiolex®, for the treatment of seizures associated with Lennox-Gastaut syndrome or Dravet syndrome in patients two years of age and older and with ongoing clinical trials in tuberous sclerosis complex.

Elizabeth A. Thiele, MD, ***TSC Champion Award***, for her passionate, unwavering clinical care for individuals with TSC throughout their lifetimes and enduring commitment to TSC-Associated Neuropsychiatric Disorders (TAND).

Dr. Thiele is Director of the Pediatric Epilepsy Program and Director of the Herscot Center for Tuberous Sclerosis Complex at Massachusetts General Hospital and a Professor of Neurology at Harvard Medical School where she directs clinical research efforts in the neurologic aspects of tuberous sclerosis complex including epilepsy, autism and mental health issues. Dr. Thiele has received numerous awards over the years, including the Manuel R. Gomez Award from the Tuberous Sclerosis Alliance in 2008, the inaugural John M. Freeman Award in 2014 and the Infantile Spasms Award from the Child Neurology Foundation in 2014. Her contributions to the medical/scientific community have been extraordinary, as demonstrated most recently with her leading the clinical trials in Epidiolex and the Epilepsy Working Group for the 2011 and 2018 TSC Clinical Consensus Guidelines updates. Dr. Thiele has illuminated the need for greater attention to TAND and has delivered compassionate care of the TSC community for more than 20 years. She was one of the first physicians to champion care for adults with TSC, particularly improved transition and adult resources.

For more information, please visit www.tsalliance.org/45Gala.

My Daughter's TSC Story of Triumphing Over Seizures: 10 Years Later

BY LISA HALL

In November 2008, my daughter Jamie Youngblood Holland's life was changed forever – in a positive, joyful way. She was a very courageous women to have made the decision at that time to undergo two brain surgeries just seven days apart, requiring 16 days in the hospital. She subsequently appeared on the cover of the Summer 2010 issue of *Perspective* after a long year of recovery. Ultimately the surgeries changed the course of her life, and I believe she is a miracle!

I now want to share an update on her illuminating story, which may shine light on someone out there facing similar TSC challenges today. I can't help but think recent research and medicine have advanced for those who endure TSC. It seems that everyone's battle is different. Ten years ago, I wrote in great detail about her battle with TSC, how it progressed throughout the years, how the anti-seizure medications could not control her seizures, and her conclusion.

I re-read the Summer 2010 article prior to writing this one. It is still very emotional as I recall her previous struggles, but let's move forward to now, 10 years later. Jamie is currently 42 years old and has never had another seizure since the surgeries. I thank God every day of my life for healing my little girl. Jamie now enjoys a successful career with the Federal government, is married, takes only two pills a day and still has no weeds in her gardens!

The healing process took many years, as the doctors said it would. Her goal was to conquer the obstacles of recovery from the brain surgeries. Jamie was very determined and strong willed with her decision to do whatever it took to overcome the "textbook" difficulties of TSC. I believe her positive outlook and attitude helped her become who she is today.


Lisa and her daughter Jamie.

Most people don't know what the hippocampus is or what it controls. Jamie's second surgery removed the left hippocampus. The absence of the left hippocampus affects memory, to name just one after-effect as there are many. Jamie's brain had to "reorganize" for the right hippocampus to take over. This simplification cannot accurately describe what this all means, how it happens or the time and effort it took for her to recover. However, Jamie's long recovery now means no more seizures and a quality of life beyond her wildest dreams. I believe God lead us to the Cleveland Clinic and to gifted neurosurgeons who gave Jamie the fortitude to rally. She now lives life to the fullest.

Jamie is very active. Her energy is endless. Without needing large amounts of medications at this point, there is no more "fog." She excels at work, is a member of the ladies' auxiliary at the local fire house, goes to the beach, helps others out of the kindness of her heart, spends time with family and friends, and loves country music line dancing. Years ago, I used to have recurring dreams that she would drive me places. I felt bad thinking that may not be possible, but it happens all the time now! She drives me around when we do things together. It is still so exciting to this day.

I'd like to share one example of her sincere kindness. For many years Jamie could not drive and needed help getting to and from work. I drove her most of the time, but there were circumstances when I was unable to do so. Many others helped by providing transportation in my absence. Several years after Jamie was healed and had been driving for quite a while, she met a co-worker in her department who could not drive. Her co-worker lived pretty close to her home, so she would pick him up every day to take him to and from work. She paid it forward! Jamie has abundant compassion for others.

I am so proud of Jamie although "proud" does not totally define how we all feel about her perseverance. She never gave up. There were many, many difficult obstacles to cope with and overcome. Resigning from society was never an option, not even a thought. When she was only a few years old, learning to talk, if it was time to get dressed or brush her hair (something "mommy" took care of), she would say, "I do it . . . I do it!" As you can tell, she was determined from the start. Jamie's great grandmother always got a chuckle from her innocent, but serious words. On many occasions throughout life, the three of us would recall those words: "I DO IT!"

I'm so glad to share a happy outcome about someone living with TSC. The genetic part remains, but the seizures are gone. I can't believe 10 years have passed. The memories are very vivid for me what Jamie endured, including the years before and after her healing. Her light shines very bright. She is living proof that miracles are real!

TSC Community Needs and Priorities Shape 2019-2023 Strategic Plan

BY REBECCA ANHANG PRICE, PHD

Throughout 2018, a Strategic Planning Committee worked closely with the TS Alliance staff and Board of Directors to update the organization's strategic plan for 2019 – 2023. Committee members represented a wide spectrum of the TSC community, as well as clinicians and researchers.

At the core of the strategic plan is understanding the needs, concerns and priorities of the TSC community. To gain this understanding, we sought input from community volunteers at the Volunteer Leadership Summit in March 2018 and conducted an online Constituent Survey from mid-January through mid-March. We are thankful to Henry Shapiro, Corporate Advisory Board Member and former Board Chair, who provided in-kind support from his company MarketCast for surveying and tabulation, and to Reiko Donato and Nan Martin, who donated their time to analyze and report survey results.

Nearly 500 people age 13 years or older responded to the survey, including individuals with TSC, parents or other primary caregivers of people with TSC, and researchers and medical practitioners. The survey was closely modeled after the Constituency Survey the TS Alliance completed in 2010, so we could look at trends over time. Survey responses reflect the opinions and experiences of those who volunteered to complete the survey online. Figure 1 shows some highlights of what we heard.

The Greatest Challenges

When asked about the TSC-related ***medical issues*** that were hardest to manage, individuals with TSC and their caregivers were most likely to mention intellectual disability and developmental delays, learning disabilities, autism spectrum disorder, behavior problems, psychiatric problems, and epilepsy.

FIGURE 1: Challenge of Managing Medical Symptoms by Percent Rated Extremely/Very Difficult to Manage

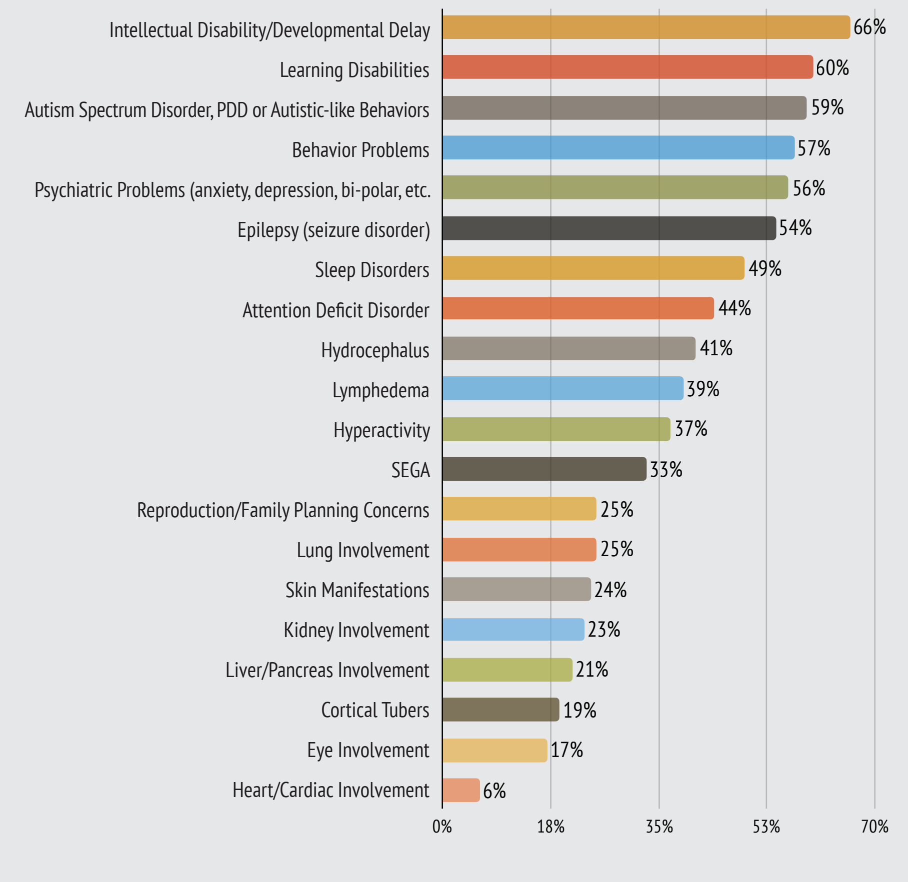
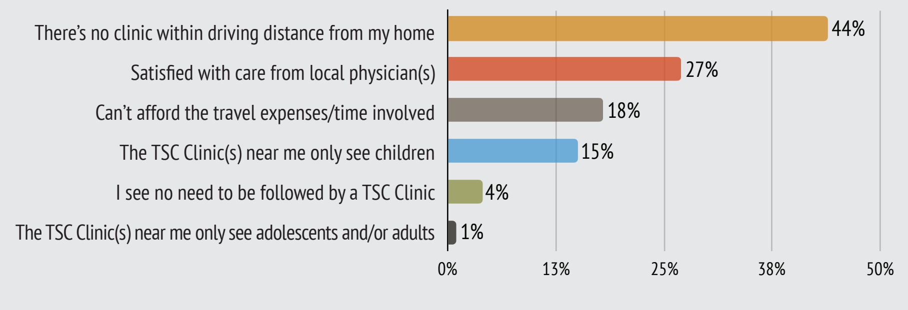


FIGURE 2: Reasons for Not Visiting TSC Clinic by Percentage



When asked about the TSC-related ***non-medical issues*** they'd ever experienced, nearly three-quarters (74%) of individuals with TSC and their caregivers mentioned education and about half mentioned obtaining and maintaining affordable health insurance coverage (52%) and accessing social and recreational programs (49%).

Getting Health Care for TSC

Four out of five (81%) individuals with TSC and their caregivers are satisfied with the care they receive, but about half find it hard to find TSC specialists near home (52%) and a third don't have access to the providers necessary to manage TSC symptoms (33%).

FIGURE 3: Ratings of TS Alliance on Selected Services by Percent Rated Excellent or Very Good

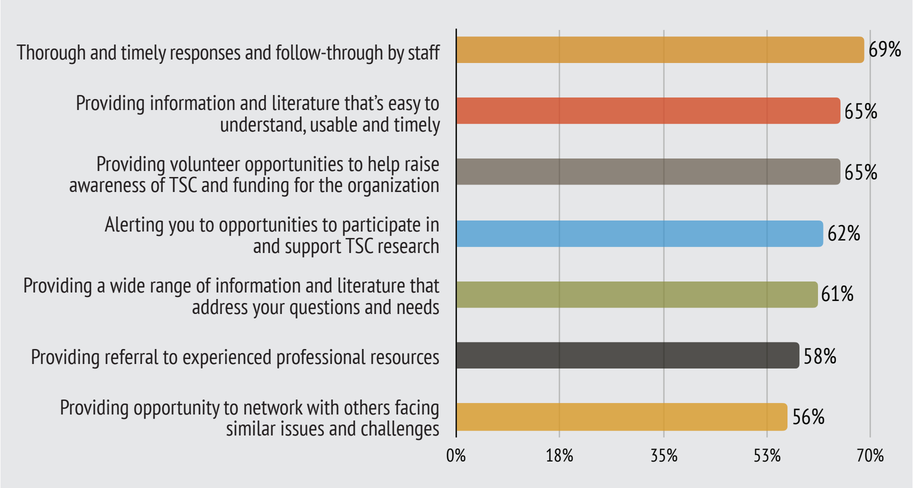
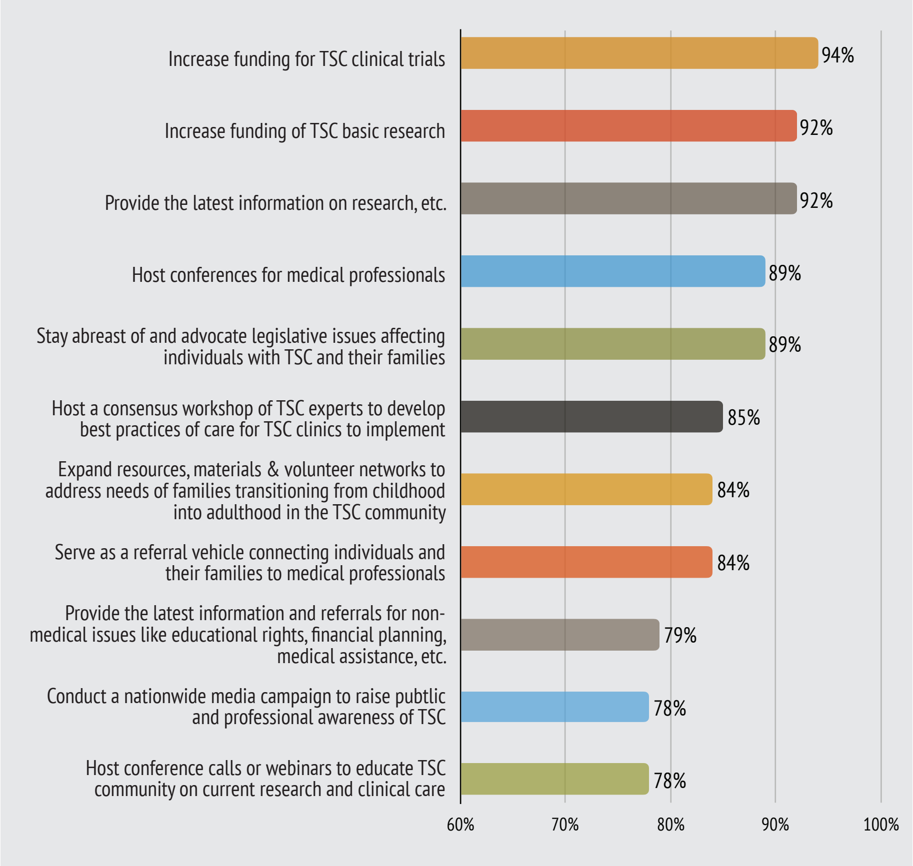


FIGURE 4: Activities to Pursue by Percent Rated Extremely or Very Important



Two-thirds of survey respondents indicated that they visit a TSC clinic. Of those who do NOT, 44% say it's because there's no clinic within driving distance of home (see figure 2).

How Well TS Alliance Meets Needs

The most highly rated TS Alliance services were thorough and timely responses and follow-through by staff (69%) and information and literature that's easy to understand, usable, and timely (65%). Survey respondents indicated more room for improvement in providing referrals to experienced professional resources and providing opportunities to network with others facing similar issues and challenges (see figure 3).

What TS Alliance Should Do Next

Activities that respondents indicated were extremely or very important for the TS Alliance to pursue were increasing funding for TSC clinical trials (94%), basic research (92%), and providing the latest information on research (92%). See figure 4.

The Strategic Plan is summarized in an "OGSM" document that describes the overall Objective of the TS Alliance's work (the O in OGSM), its Goals (G), Strategies to achieve those goals (S), and Measures to allow the Alliance to track performance on each goal (M). The growing importance of access to medicines and high-quality TSC care led to the inclusion of a new Goal for 2019 – 2023, "Improve Access and Quality of Care." You can view the entire OGSM at www.tsalliance.org/OGSM.

Strategic Planning Committee Members

- Rebecca Anhang Price, PhD, *Chair*
- Martina Bebin, MD, MPA
- Matt Bolger
- Marc Cohen, MD, MPH
- David Coit
- David Fitzmaurice
- Rob Grandia
- Heather Lens
- Cathy Krinsky
- Darcy Krueger, MD, PhD
- Tara Zimmerman

Focus on School Challenges: The Road Less Traveled

BY JULIE COMEAU

EDUCATION PARENT MENTOR, LOUISIANA DEPENDENT ADULT TRANSITION RESOURCE COORDINATOR

We are all on the road less traveled. I used to find myself measuring the progression of my journey by the days before tuberous sclerosis complex (TSC), and the days since. I have drifted deep into the thick woods, traveling alone at times. I have questioned doctors, and I have questioned my faith. Then there are those moments representing roses along the path. You stop and savor their beauty and aroma, despite the thorns that are one day certain to make you bleed with fear and pain. Every milestone reached was a treasure to be admired. A baby's smile never looked more beautiful. I always seemed to follow the roses back to my path of hope, and they seemed to arrive at a time when needed most. Still, I doubted myself. My biggest fear was I wouldn't see the pain of the struggle. I would become numb, and that would become my normal. Would I recognize that I was stuck? Then my starting point for my journey changed. My measure of what was traumatic changed. It was the moment blind faith and trust in education was shattered, and I was stuck.

I found myself far from familiar trails as my son, Matthew, transitioned to middle school. We were already experiencing some of the behaviors associated with TSC-Associated Neuropsychiatric Disorder (TAND). There were multiple occasions I had requested a behavior intervention plan at his school and without that support, Matthew was subjected to repeated undocumented removals. They continued until the day he finally told me what was happening. Without current training in TSC and epilepsy, the school frequently claimed he was "faking" seizures or using them to manipulate. With ineffective supports and reluctant staff, the frequency of behaviors increased and intensified. There is very little doubt in my mind he felt threatened and unsafe. It was impossible for me to work. I was often called to the school for seizures and behaviors, until one day I arrived to find him lying motionless on the floor.

The path of my journey suddenly changed. I was no longer walking a difficult path – I was in a boat, and seas were rough. Water was slowly sinking my ship. I paddled hard through every wave, including police reports, state complaints, Office of Civil Rights complaints, and due processes. I filed due process seeking staff training. One year later, the school district filed against me seeking a vocational assessment and specific medical records. I was being "forced" to void my son's right to privacy and HIPPA protections of his medical records and voiding my right to voluntary consent. I did not agree to this resolution. I was weary from three years of paddling, and I wanted out of this sinking ship. I was having unprovoked panic attacks.

In June 2016, I filed a motion to dismiss due process. I withdrew Matthew from school, and we carved our way and found his path to healing. He improved quickly at home. The once daily non-epileptic seizures grew farther and farther apart. The volatile


Matthew and Julie Comeaux.


Matthew Comeaux.

moods and destructive behaviors responded well as we implemented *Collaborative & Proactive Solutions* by Dr. Ross Greene. It took time, but he was once again embracing academics. I was out of that boat and finally seeing the light through the forest. His psychiatrist was also impressed with his progress. I was finally letting my guard down and admiring the roses again, until another thorn dug in deep.

On December 30, 2017, Matthew had flashbacks to the day he describes as people holding him down. We found ourselves having to restrain him in the yard, calling for an ambulance as he looked at us with fear. He called out, only he wasn't speaking to or seeing us. He was repeatedly pleading with "Mr. L" to let go and don't hurt his arm. Tears burned my eyes and streamed down my cheeks, and that thorn went straight for my heart. My heart believed I failed to protect him from a pain no child should ever know at school, and this pain had a deeper hold on him than I could have ever imagined.

Our journey has been challenging and frightening, but I have learned I have a fire and a passion for building strong parents to become diplomatic advocates for their children. I have embraced this opportunity to volunteer for the Parent Education Mentor program with the Tuberous Sclerosis Alliance. We have to fight and sacrifice so much for our children. We have spent sleepless nights in the NICU, in the E.R., and at home. Education is a scary hurdle, but there is great value in having the tools needed to have your child succeed, and it is always a less frightening journey when we have the comfort of someone walking that weary road, holding our hand.

MANUEL R. GOMEZ AWARD NOMINATIONS NOW BEING ACCEPTED

The Tuberous Sclerosis Alliance grants the Manuel R. Gomez Award to individuals who have made a significant impact on our understanding of TSC in research, and/or have impacted the delivery of clinical care for individuals with TSC. The award is given in memory of Manuel R. Gomez, MD (1928-2006), the "Father of TSC" in the United States. A dedicated physician-scientist, Dr. Gomez provided care and compassion for hundreds of individuals with TSC while conducting some of the pioneering research in the field.

To nominate someone for the Manuel R. Gomez Award, please prepare a letter that describes how the nominee has contributed to clinical practice and/or research in the TSC field. Please send the letter and the nominee's curriculum vitae by email to cho@tsalliance.org or by mail to Tuberous Sclerosis Alliance, Attn: Calvin Ho, 801 Roeder Road, Suite 750, Silver Spring, MD 20910. Applications are accepted until April 12, 2019.

Previous Gomez Award Recipients

- **Mark Nellist, PhD**
  Erasmus MC
- **Chris Kingswood MB, BS, FRCP**
  Brighton and Sussex University Hospitals NHS Trust
- **Julian Sampson, DM, FRCP, FMedSci**
  Cardiff University
- **Paolo Curatolo, MD**
  Tor Vergata University Hospital of Rome
- **Petrus J. de Vries, MBChB, MRCPsych, PhD**
  University of Cape Town
- **John J. Bissler, MD**
  University of Tennessee Health Science Center
- **E. Martina Bebin, MD, MPA**
  University of Alabama at Birmingham
- **Howard Weiner, MD**
  Texas Children's Hospital (formerly at New York University Medical Center)
- **Mark Mausner, MD**
- **Sergiusz Jóźwiak, MD, PhD**
  Children's Memorial Health Institute
- **Elizabeth A. Thiele, MD, PhD**
  Carol and James Herscot Center for Children and Adults with TSC at Massachusetts General Hospital
- **Ann Hunt**
  Tuberous Sclerosis Association
- **David Neal Franz, MD**
  Cincinnati Children's Hospital Medical Center
- **Elizabeth Petri Henske, MD**
  Brigham and Women's Hospital (formerly at Fox Chase Cancer Center)
- **Hope Northrup, MD**
  University of Texas Health Science Center at Houston
- **E. Steve Roach, MD**
  The Ohio State University College of Medicine (formerly at Wake Forest University)
- **David Kwiatkowski, MD, PhD**
  Brigham & Women's Hospital
- **Vicky Whittemore, PhD**
  National Institute of Neurological Disorders and Stroke (formerly at the Tuberous Sclerosis Alliance)

My Family's Journey with TSC

By Kimberly Brenton

Kimberly and Meagen Brenton.

Let me introduce myself. My name is Kimberly Brenton. I live with my daughter, Meagen, and my husband, Meagen's step father, in Ellsworth, Maine.

My journey with tuberous sclerosis complex (TSC) began later in life than many of my fellow friends with TSC. I was 31 when diagnosed with TSC. Meagen started to show signs of seizure activity in late Summer 2003. She was not even a year old yet.

I unfortunately knew what a seizure looked like. My mom suffered from seizures. She started to experience them at 13 years old but hasn't had a seizure since her early 30s. She too was clinically diagnosed with TSC. I never have experienced a seizure myself. I have my suspicions that TSC came down my maternal grandmother's side, but she and her siblings had passed before our TSC diagnoses.

We were referred to an amazing pediatric neurologist soon after we visited our family doctor. Dr. Wendy Stewart saw Meagen for the first time. Fortunately, Meagen had a seizure during the appointment. It gave Dr. Stewart a better understanding of the type of seizures she was having, which was crucial in treatment. Throughout the appointment, Dr. Stewart began to gather an extensive health history for me and Meagen. She ruled out Meagen's seizure as being fever induced or caused by falling and hitting her head.

She also noted the birthmark on my lower back, a Shagreen patch, and fibromas under my toe and fingernails. Dr. Stewart concluded that she thought our diagnosis was indeed TSC. She wrote the three words on the back of appointment card, "tuberous sclerosis complex." She said she would order more testing for both Meagen and me to determine if TSC was the correct diagnosis.

Those three words changed our lives going forward. I wasn't scared, I just wanted Meagen to feel better and have no more seizures. On December 31, 2003, I had a CT of my brain and abdomen. Meagen already had extensive testing. A DNA test was also ordered for both of us, and the results confirmed we both had TSC. The official result didn't come back to our doctors until March 2004.

Despite medication, Meagen continued to have seizures. Howver, she was still meeting all her milestones and was determined to continue to advance until she was 18 months old when her vocabulary decreased from about 100 words to around 25. I was then introduced to the world of Speech Therapy, Occupational Therapy, and Physical Therapy. Meagen was enrolled into a developmental preschool as well at this time.

While Meagen endured seizures, and I ran around getting her to all her appointments, my CT scan results showed a 5.3 cm mass on my right kidney. This development was just another cog in the perpetual wheel in the TSC world that soon became our "normal."

Searching for a kidney specialist that would see me was hard. In Maine, the medical community in my area was not familiar with TSC or confident to treat me. We also were fighting insurance coverage to see specialists out of state. We finally found a doctor in the southern part of the state who would see both of us – Dr. Wendy Smith, a genetics specialist. Dr. Smith was able to coordinate a nephrologist with knowledge of TSC who would actually see me and discuss any possible treatments. With the recommendations from Drs. Stewart and Smith, our family doctor and my nephrologist, our insurance company finally approved an appointment with Dr. Elizabeth Thiele at The Carol and James Herscot Center for Children and Adults with TSC at Massachusetts General Hospital in Boston.

Without these people over the past 16 years, I could not have navigated the world of TSC as well as I have. I hope sharing my family's story has given you hope and let you know you aren't alone.

Before Meagen's first appointment with Dr. Thiele, I started seeing my nephrologist, who recommended embolization as a treatment for me. Then, in September 2004, I made an appointment for Meagen with our family doctor for suspected strep throat. As we were leaving, the doctor said, "Oh, by the way, I heard from your nephrologist, and he thinks you may have cancer." He tried to assure me it was a long shot but said I should call the nephrologist.

This development started a whole new chapter in my TSC journey. My nephrologist consulted a local urologist, and they concluded an embolization was not the solution and suggested I have the mass and right kidney removed. Given that information I didn't wait; I scheduled the surgery for December 22, 2004.

On December 10, 2004, Meagen and I saw Dr. Thiele. She was concerned about me having the surgery, because I could potentially remove a perfectly healthy kidney despite the mass. I let her know I was committed and knew it was the right thing to do, because deep down I knew it was not good and needed to be taken care of as soon as possible.

My nephrectomy was a success with no complications. As the anesthesia kicked in, I remember someone in the OR said, "Wow, we don't usually see such a healthy patient for this type of surgery." The next thing I knew they were waking me up in recovery. Looking back on this now I should have been scared, but I wasn't – probably because I was so involved in keeping Meagen safe and making sure she was getting everything she needed.

Following the surgery, the doctor came in a short time after I was taken to my room. He said, "We got it (the cancer) just in time." The mass was measured 6 cm plus, and the cancer was contained within the vascular mass and kidney. I was fortunate not to need addtional cancer treatments.

Looking back, if it weren't for Meagen having those first seizures, I don't want to think where I would be, because I had no symptoms associated with kidney involvement,or function except borderline high blood pressure. There just wasn't a reason for a doctor to check my kidney function. Meagen saved my life. She is my angel.

Our adventure didn't end with my surgery. A few months later, Meagen underwent an evaluation for brain surgery, since medication wasn't stopping or slowing her seizure activity. There were many trips to Boston for testing and many lengthy stays at "Hotel Mass General" as we fondly called it. These trips became our new normal.

In November 2005, Meagen was deemed a candidate for brain surgery. The surgery was scheduled for December 8, 2005, coincidently the day before her third birthday. The surgery went well. Our stay was lengthened because Meagen developed a virus that quarantined us. Meagen was discharged on December 17, 2005. Meagen ultimately had a limited frontal lobe resection. She recovered quickly, and it was like having a different child. She was more alert and went medication and seizure free for a little over two years. Our new normal yet again changed.

Meagen's seizures came back and medication was started again. Our adventure with TSC was like riding a roller coaster. The emotions, the appointments and the seizures were up and down. I learned to advocate for Meagen and myself. I questioned what didn't make sense, and most importantly I knew others with TSC weren't as fortunate just to deal with these minor things. Their normal was much more involved than our's.

November 16, 2012 was the last day we witnessed a seizure. Meagen has been seizure free for six years. In September 2011 a pancreatic lesion was found near her spleen; a biopsy showed it was likely to contain cancerous cells. On December 2, 2011, the lesion was removed. Meagen recovered quickly and returned to school after the Christmas holiday. We actually made it out of the hospital before her birthday.

But TSC wasn't done messing with us. In 2015, Meagen was scheduled for another pancreatic surgery. On August 28, 2015, a neuroendocrine tumor was removed containing abnormal (i.e., cancerous) cells replicating at a 3% rate. Meagen was bound and determined not to miss the first day of school the following Tuesday. She was discharged on Sunday, and she did indeed start school with her friends on the first day of 7th grade. Her only disappointment was not being able to go to PE for a month.

Now a high school sophomore and still seizure free, Meagen is progressing and works hard. She understands she may have to study harder than her peers. I tell her if she is trying her best then there won't be a problem.

We are currently stable in our TSC journey. I have been active in the TSC community, trying to share my experiences and knowledge. I provide "my story" in a written format with the awesome volunteers who March on Capitol Hill every year for Federal TSC research funding. I believe that without this funding, Meagen would not be seizure free, and I probably wouldn't be here. I still meet medical professionals who do not know about TSC, and I know that by sharing my medical history and what TSC is, I have made a significant difference.

This year Meagen and I were fortunate to be able to attend the 2018 World TSC Conference in Dallas. It was an experience unlike anything we have participated in before. It showed Meagen there were other teenagers with TSC like herself. I learned a lot and met many people I had communicated with through email, social media and phone calls. These people and many others who didn't attend have affected my life in such a positive and caring way.

Without these people over the past 16 years, I could not have navigated the world of TSC as well as I have. I hope sharing my family's story has given you hope and let you know you aren't alone. While I may not know what TSC will throw at us in the future, I do know I have TSC, but TSC doesn't have me.

Honorariums (August - November 2018)

You can honor a friend or family member for an important occasion with a gift to the TS Alliance. It is a wonderful way to send a birthday or anniversary wish, or congratulations for retirement, a job well done, graduation, etc. Please include the name and address of the individual being honored so that acknowledgement of your kind donation can be sent. Tuberous Sclerosis Alliance honorarium cards are also available if you would like to make a gift in honor of family, friends, or colleagues. To receive tribute cards, contact Justin Martucci at (240) 638 4643 or jmartucci@tsalliance.org.

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Memorials (August - November 2018)

Contributions are given to the Tuberous Sclerosis Alliance at the request of family members in memory of their loved ones. We extend our sympathies to the family and friends of those memorialized below. These generous contributions support the progress of our mission to find a cure for tuberous sclerosis complex.

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Tribute(s) for W.L. Tew
Ms. Susan Jones
Tribute(s) for Brittany Webb
Ms. Toni Y. Brown
Tribute(s) for Christopher T. Winkler
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SEE THE IMPACT OF YOUR GENEROSITY TODAY

Make a Tax-Free Gift From Your IRA

As you may already know, the Protecting Americans from Tax Hikes (PATH) Act, popularly known as the IRA charitable rollover, has been made permanent. If you are 70 ½ years old or older, you can take advantage of a simple way to help those we serve and receive tax benefits in return. You can give any amount up to $100,000 per year from your IRA directly to a qualified charity such as ours without having to pay income taxes on the money. This law no longer has any expiration date so you are free to make annual gifts to the Tuberous Sclerosis Alliance Endowment Fund (TS Alliance Endowment Fund) this year and well into the future.

Why Consider This Gift?

Your gift will be put to use today, allowing you to see the difference your donation is making.

You pay no income taxes on the gift. The transfer generates neither taxable income nor a tax deduction, so you benefit even if you do not itemize your tax deductions.

If you have not yet taken your required minimum distribution for the year, your IRA charitable rollover gift can satisfy all or part of that requirement.

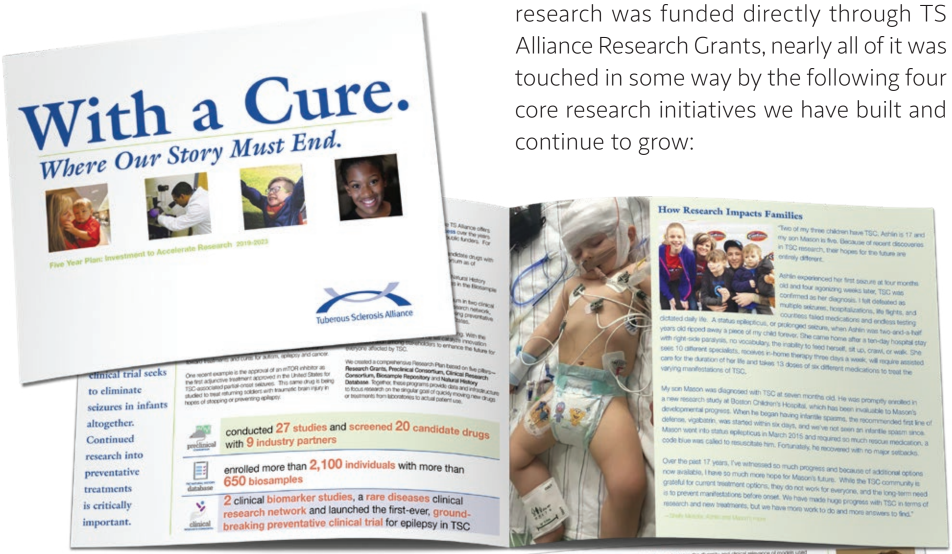
If you want to make a qualifying transfer, contact your IRA administrator and instruct them to transfer a contribution to the TS Alliance Endowment Fund.

To learn more about using your IRA to make an impact at TS Alliance Endowment Fund, contact Lisa Moss at (301) 562-9890 or (800) 225-6872 or lmoss@tsallinace.org.

TS Alliance Unveils Comprehensive 5-Year Research Business Plan

The TS Alliance plans to raise $56 million by 2023 to speed up TSC research. Building on the depth of our relationships with both the TSC and research communities, the TS Alliance developed a *Five Year Plan: Investment to Accelerate Research* to create unparalleled resources that will help scientists find a cure for this devastating disease. For more detailed information, you can download the plan's *Executive Summary* at www.tsalliance.org/ExecSummary.

Our plan aims to alter the course of the disease, improve the quality of life for those living with TSC and create a more hopeful future for those who will be born with TSC. The funds we raise will be used to stimulate and encourage research on new therapies. Moreover, the plan will strengthen and expand the reach of our four core research programs, which span the entire pipeline of development for new drugs and other treatments.



With a Cure.

Where Our Story Must End.

Five Year Plan: Investment to Accelerate Research 2019-2023

clinical trial seeks to eliminate seizures in infants altogether. Continued research into preventative treatments is critically important.

conducted 27 studies and screened 20 candidate drugs with 9 industry partners

enrolled more than 2,100 individuals with more than 650 biosamples

2 clinical biomarker studies, a rare diseases clinical research network and launched the first-ever, ground-breaking preventative clinical trial for epilepsy in TSC

How Research Impacts Families

What Our Research Investments Have Accomplished So Far

When the TS Alliance gave its first research grant in 1984, the future for people affected by TSC was bleak. Thankfully, the outlook has changed dramatically. Through tireless research in laboratories and clinics, scientists have determined how mutations in one of two genes can cause the manifestations of TSC. They've built on that knowledge to create safe and effective drugs specifically for TSC.

While more than $20 million of this research was funded directly through TS Alliance Research Grants, nearly all of it was touched in some way by the following four core research initiatives we have built and continue to grow:

- **Innovative Research:** Our Research Grants have encouraged young scientists to study TSC and helped to generate new hypotheses about how to treat the disease. Our biennial International Research Conferences have led to new collaborations among the world's leading experts on TSC.
- **Natural History Database and Biosample Repository:** Pairing information about an individual's TSC experience with one or more biological samples allows scientists to figure out how and why TSC varies so much from person to person.
- **Preclinical Consortium:** Creating robust and standardized ways to test new drugs encourages both academic and industry researchers to try new ideas for treating TSC.
- **Clinical Research Consortium:** Making it easier to execute clinical trials in TSC means we can learn much more quickly if new therapies work. Developing consensus guidelines and evidence-based practices helps all people with TSC receive the best medical care possible, no matter where they live or which doctor they see.

Where We Need to Go Now

Though we have come far since 1984, much work remains. For instance, why are some people with TSC at higher risk for certain manifestations, such as seizures? If we can figure that out, can we prevent seizures in susceptible individuals before they ever occur? Can we find drugs that have fewer side effects than the ones on the market today? And can we find better treatments for TSC-Associated Neuropsychiatric Disorders (TAND)?

We are very close to having answers to these questions. To reach the finish line, we need to speed the process of turning new scientific findings into a drug, treatment or screening tool available to the community, which is exactly why we are implementing the *Five Year Plan: Investment to Accelerate Research*.

The *Five Year Plan* builds on our four core research initiatives to bring high priority treatments into the drug development pipeline and encourage academics and companies to develop new therapies for TSC.

Bringing Promising Drugs to Clinical Trials and Improving Clinical Care

The mission of the TS Alliance is to find a cure for TSC, while improving the lives of those affected. All potential drugs to treat TSC must go through clinical trials to prove they are safe to take and effective against the aspect of the disease they are targeting.

The Clinical Research Consortium was formed to make it easier and more successful to run clinical trials in TSC. Since then, the consortium has grown to 15 research sites across the country and has been awarded more than $29 million from the National Institutes of Health (NIH) to carry out four trials and studies. It is currently running the first-ever clinical trial in the United States aimed at preventing epilepsy in infants with TSC.

The *Five Year Plan* will allow us to increase the impact of the Clinical Research Consortium even further. We plan to establish a Clinical Research Acceleration Fund to cover unanticipated costs that would otherwise slow clinical research. This initiative will include travel funding to enable people with TSC to participate in trials, even if they do not live near one of the consortium sites, and support for clinical research coordinators to put more of their valuable time and effort into TSC trials.

With the *Five Year Plan*, we also plan to improve clinical care for people with TSC by building a system that helps TSC Clinics share data and develop best practices based on real-world evidence. The evidence will help us develop guidelines for TSC care we can share with all healthcare providers to help us ensure everyone with TSC has access to the best care possible. Additionally, our vision is for TSC to be included in the newborn screening program by 2023 so every child with TSC is diagnosed and treated as soon as possible.

Testing New Drugs to Treat TSC

In order to take a drug to clinical trial, researchers need strong evidence it works in a cell or animal model of the disease, which is why the TS Alliance established the Preclinical Consortium in 2016. This consortium allows researchers to test new drugs in a consistent, rigorous and reproducible manner, using the best available animal models of important TSC manifestations.

Under the *Five Year Plan*, the TS Alliance will increase the Preclinical Consortium's capacity to test new drugs, with the goal of testing 75 new candidate drugs or combinations by 2023. We will also develop the ability to find new potential drugs through rapid screening with cell-based tests. To help early-stage startup companies with promising ideas, we will use venture philanthropy to help them access this consortium's resources.

> *Our plan is ambitious because we know how important it is to develop new treatments and improve the lives of people living with TSC. You can help us reach these goals by participating in our programs.*

We also want to ensure the Preclinical Consortium's work addresses the TSC community's most pressing needs. To do so, we are investing in the creation and evaluation of animal and cell models that can help us test which mechanisms might work to treat TSC-Associated Neuropsychiatric Disorders, epilepsy and tumors.

Understanding How TSC Affects You

We cannot develop new therapies for TSC without understanding how TSC affects individuals as they get older and how they respond to currently available treatments. Our Natural History Database and Biosample Repository pair detailed medical data with blood, DNA and tissues to help researchers determine how and why TSC varies from person to person.

Currently, only people who are seen at a handful of TSC Clinics can participate in these projects. The *Five Year Plan* will allow us to open up participation to everyone with TSC. We also plan to allow individuals and caregivers to contribute information about how they feel and function directly into the Natural History Database. To make it easier for researchers to use these data and samples, we intend to analyze all biosamples centrally and dedicate a portion of our research grant funding for projects using biosamples and natural history data.

Stimulating Innovative TSC Research

None of this can happen if scientists are working in their own bubbles, unaware of what other researchers are doing and what matters most to the TSC community. To fill this gap, the TS Alliance holds an International TSC Research Conference every other year. The *Five Year Plan* allows us to complement those large gatherings with smaller Innovation Workshops once or twice a year focused on the highest-priority research needs. In addition, a new Innovation Fund will allow us to fund high priority projects as quickly as possible.

Help Us Move Forward

Our plan is ambitious because we know how important it is to develop new treatments and improve the lives of people living with TSC. You can help us reach these goals by participating in our programs. The Clinical Research Consortium needs more people with TSC to enroll in clinical trials, so we can figure out whether new treatments will be safe and effective. The Natural History Database and Biosample Repository need to enroll more participants, so researchers can have more data and samples to work with. Finally, local Step Forward to Cure TSC walks, the upcoming 45th Anniversary Gala and other fundraising programs directly support all four major research initiatives. We hope you'll join us today to help accelerate research in TSC.

If you'd like to receive a full version of the *Five Year Plan: Investment to Accelerate Research* or learn more about how you can invest in our future, please contact President & CEO Kari Luther Rosbeck at ksrosbeck@tsalliance.org or Senior Director of Donor Relations Lisa Moss at lmoss@tsalliance.org. You may also call them at (301) 562-9890.

TSC Clinical Trials

The following clinical trials are currently recruiting participants. If you are interested, please use the contacts listed in each trial description:

Preventing Epilepsy Using Vigabatrin in Infants with Tuberous Sclerosis Complex (PREVeNT Trial)

Where: TSC Clinical Research Consortium sites at Boston Children's Hospital, Cincinnati Children's Hospital Medical Center, University of Alabama at Birmingham, University of California at Los Angeles, University of Texas at Houston, Minnesota Epilepsy Group, PA, Stanford University, Beaumont Hospital, Washington University in St. Louis, Children's Hospital Orange County (CHOC), Children's Hospital of Philadelphia (CHOP), and Duke University Medical Center.

Principal Investigators: Mustafa Sahin, MD, PhD (Boston), Darcy Krueger, MD, PhD (Cincinnati), Martina Bebin, MD, MPA (Birmingham), Joyce Wu, MD (Los Angeles), Mary Kay Koenig, MD (Houston), Michael Frost, MD (Minnesota), Brenda Porter, MD, PhD (Stanford), Danielle Nolan, MD (Royal Oak, MI), Mary Zupanc, MD (Orange, CA), Katherine Taub, MD (Philadelphia), Klaus Werner, MD, PhD (Durham, NC).

Who is eligible to participate? We are enrolling 0- to 6-month old infants with a diagnosis of tuberous sclerosis complex (TSC) and no history of seizures for a new study on prevention of epilepsy. The goal of this project is to use EEG, behavioral testing and early use of vigabatrin to help determine the developmental impact of epilepsy from birth to 36 months of age.

What will we do? If you agree to participate, the research team will obtain your informed consent. The study involves up to 13 visits over a three-year period. The study visits will include behavioral testing, EEGs, eye exams, and physical exams, optional blood draws.

Cost/time commitment: Study visits will vary in length based on your child's age, but generally be a few hours in time. There is no fee to participate in this study. Travel funding may be available.

Results: Summary scores of your child's behavioral testing and EEG results will be provided to you. Every step of the way, if new results from the testing are concerning, we will notify you and assist you in obtaining referrals or interventions. After all study data has been analyzed, we will inform families of the overall results.

Contact: If you are interested in learning more, contact Regina Ryan, PREVeNT Project Manager, at prevent@uabmc.edu or (205) 975-2890.

JASPER Early intervention for Tuberous Sclerosis Complex

Researchers from Boston and Los Angeles are studying the benefits of early behavioral intervention in infants and toddlers with TSC. Intervention is provided in person and now remotely via video conferencing. This study design allows us to include families from all over the country.

Where: Assessments will take place at UCLA and Boston Children's Hospital. 4 intervention sessions will take place at the study sites, the rest will occur in your home, through video conferencing.

Principal Investigators: This NIH funded study is led by Shafali Jeste, MD. (UCLA), Connie Kasari, Ph.D. (UCLA), Charles A. Nelson, Ph.D. (Boston Children's Hospital) and Elizabeth Thiele, MD, Ph.D. (Mass General Hospital)

Who is eligible to participate? Any child with a diagnosis of TSC between the ages of 12-36 months is eligible for the intervention. Children below 57 months may also be eligible for one-time participation in the study.

To learn more about the study, visit www.JETSstudy.org or contact Carly Hyde/UCLA at chyde@mednet.ucla.edu or 310-825-8738, or Jack Keller/Boston Children's at tscjasper@childrens.harvard.edu or (857) 218-3010.

Clinical Trial for Individuals Diagnosed with Tuberous Sclerosis Complex (TSC) and Cutaneous Angiofibromas

Researchers are currently conducting a clinical trial for individuals diagnosed with TSC with facial bumps called cutaneous angiofibromas. The goal of this study is to investigate if the study drug, sirolimus, improves cutaneous angiofibromas in individuals diagnosed with TSC. The trial is sponsored by a pharmaceutical company named Aucta Pharmaceuticals, Inc, and it will involve 3 to 5 centers including Boston Children's Hospital and Children's Hospital of Philadelphia.

They are looking to recruit individuals between the ages of 2 and 18 years who have TSC with cutaneous angiofibromas. If you/your child qualifies for the treatment phase of the trial after the initial screening visit, you/your child will be asked to take either the study ointment or a placebo (ointment with no medicine), which is determined by chance.

The study involves 6 visits to the hospital over a 4-month (16-week) timeframe. After completing the first phase of the study, you/your child will be offered the opportunity to participate in an optional 12-week open-label phase with sirolimus ointment. If you/your child agrees to participate in the open-label phase, you/your child will make 3 additional clinic visits bringing the total clinic visits to 9.

The study visits will involve blood draws, skin exams, vital signs, laboratory tests and other evaluations such as questionnaires and assessments. The study drug will be provided at no charge, and a voucher for parking will be provided to you at each study visit.

The following study sites are currently recruiting study participants. If you are interested in participating in this trial, please contact the following study coordinator and they will assist you with any questions you may have:

- Boston Children's Hospital: Julia Costantini; (617) 355-1494; Julia.Costantini@childrens.harvard.edu
- Children's Hospital of Philadelphia: Donnette Paris; (267) 426-7167; PARIS@email.chop.edu
- Le Bonheur Children's Hospital: Tracee Ridley-Pryor; (901) 287-5338; tracee.ridley-pryor@lebonheur.org

Phenotypic Variability in Tuberous Sclerosis Complex

Dr. Narayanan and colleagues at the Translational Genomics Research Institute (TGen) in Phoenix, Arizona are conducting a research study aimed at identifying genetic modifiers of disease severity in TSC. For this study, they are recruiting families in which a parent and child both have TSC. They are looking for those families where the parent has little or no neurological problems (mild disease) and the child has severe neurological disease (severity assessed by scoring epilepsy and cognitive symptoms). Their goal is to identify those genetic differences between affected parent and affected child that might account for difference in disease severity. This might eventually allow them to devise a blood test (genetic profile) that might be used to predict disease severity and initiate disease-modifying treatments before children develop severe symptoms.

What is involved in the study? After appropriate informed consent, participating families will provide samples (blood, and in some cases skin biopsy samples if possible), which will be studied by using the latest in genomic methods.

For more information about this clinical research study, please contact Keri Ramsey at (602) 343-8620 or Newell Belnap at (928) 369-8692.

Efficacy and Safety of Lacosamide as Adjunctive Therapy in Subjects ≥1 Month to <4 Years with Partial-onset Seizures

Description: UCB Biosciences, Inc. is conducting a clinical trial to evaluate the effectiveness and safety of lacosamide (Vimpat®) in children less than 4 years of age who have partial-onset seizures. There are study locations in Alabama, Florida, Louisiana, Nevada, New Hampshire, Ohio, Texas, and Washington.

For specific site location and study coordinator phone number contact Jo Anne Nakagawa at jnakagawa@tsalliance.org or (240) 638-4654.

A Double-Blind, Placebo-Controlled, Inpatient, Dose-Ranging Efficacy Study of Staccato Alprazolam (STAP-001) in Subjects with Epilepsy with a Predictable Seizure Pattern

Engage Therapeutics is a small biotechnology company developing a rescue inhaler for drug-resistant epilepsy. They are currently looking for persons with longer (3-5 minutes in duration), predictably patterned seizures to participate in a paid trial of the product.

For more info, visit epilepsyhealthstudy.com.

2019 REGIONAL TSC & LAM CONFERENCE SERIES

The Tuberous Sclerosis Alliance and The LAM Foundation will co-host five Regional TSC & LAM Conferences in 2019 aimed at patients and caregivers in:

- **Boston:** April 13
- **St. Louis:** June 15
- **Chicago:** September 7
- **Atlanta:** September 21
- **Los Angeles:** November 2

These free one-day conferences will feature local leading researchers and clinicians specializing in TSC and lymphangioleiomyomatosis (LAM), as well as speakers from The ARC, Child Neurology Foundation and MassMutual discussing transition issues. They will also include networking opportunities to meet other families and individuals from the area. These full-day conferences will offer a Pediatric Track (Track 1), a Transition Track (Track 2) and an Adult Track with sessions important to adults with TSC and LAM (Track 3).

Key topics will include:

- Research update, including upcoming clinical studies and trials for TSC and LAM
- Seizure types and treatments (pediatric and adult)
- Tumor growth and treatments
- Behavior and mental health
- Transitioning into adulthood
- LAM
- Genetics
- Quality of life
- Kidneys and angiomyolipomas

People in each region will gather together to attend these conferences to make stronger connections with peers, researchers and clinicians in their community. Families and individuals who live with TSC and LAM, professionals who work with them and extended supporters will use the meetings to learn more about current treatment options, basic and transitional research, and clinical trials and to spend time with other people affected by TSC and LAM. These conferences will also allow the TS Alliance and The LAM Foundation to reach newly diagnosed families, identify new constituents and provide fuller support in those communities.

The free registration will include lunch. Please note: Childcare will not be available. For more information and to register, please visit www.tsalliance.org/conferences.

TSC Clinics "Up Close"

Northern California TSC Clinic at Lucille Packard Children's Hospital, Stanford Hospital and Clinics

The TS Alliance Board of Directors recognized the Northern California TSC Clinic at Lucille Packard Children's Hospital, Stanford Hospital and Clinics in October 2013 as the fifth TSC Clinic in California. This article continues an interview series about the healthcare professionals and staff who provide care and educational support to those affected. The TSC Clinic staff includes:

Brenda E. Porter, MD, PhD, TSC Clinic Director and Associate Professor of Neurology, Stanford University School of Medicine. Director of the Electrophysiology Laboratory at Lucile Packard Children's Hospital.

Scheherazade Le, MD, TSC Clinic Co-Director and Clinical Assistant Professor of Neurology, Stanford University.

Elli Brimble, CGC, Genetic Counselor, Department of Neurology

Gerald Grant, MD, FACS, Professor and Chief of Pediatric Neurosurgery, Department of Neurosurgery

Robert T. Isom, MD, Clinical Associate Professor, Department of Nephrology

Perla Bautista Muench, BSN, RN, MSN, Outpatient Epilepsy Case Manager and TSC Clinic Coordinator

Joyce M.C. Teng, MD, PhD, Professor in Dermatology and Pediatrics, Department of Dermatology

Bridget Britt, Step Forward to Cure TSC Walk Chair of Northern California and TSC Clinic Ambassador


Dr. Scheherazade Le


Elli Brimble


Dr. Brenda Porter with Molly Britt.


Dr. Robert Isom


Dr. Joyce Teng


Left to right: Kristen Yeom, MD (neuroradiologist), Gerald Grant, MD, Joanne Orozco, RN (epilepsy nurse coordinator), Eli Brimble, CGC, Perla Bautista Muench, BSN, RN, MSN.


Dr. Gerald Grant

Tell us what motivated you to choose your career path.

Dr. Porter: I worked in a research lab in high school. My mother was a laboratory technologist in the lab, and she got me the job. I would go to the medical library and use a Xerox machine to make copies of research articles. I would read them while I copied them and pretty soon I was hooked. I got my name on a research paper when I worked on a project in the lab as a high school student; I owe a big thanks to an MD, PhD post-doctoral fellow in the lab, William Roth, as he realized I had a passion for science. He took me under his wing and convinced me I could go to medical school one day and I did. I went the Medical Scientist Training Program at Washington University in St. Louis where I met my husband, so I am grateful to Dr. Roth for having faith in me. Once I discovered how cool and complicated the brain was, I knew I wanted to be a neurologist and a neuroscience researcher. I have tried hard to always think of my job as a combination of scientist, teacher and a clinician.

Dr. Le: My fascination with the brain led me to pursue neurosciences. My desire to help patients affected by nervous system disorders at the bedside led me to become an epileptologist.

Elli: I wanted an opportunity to work closely with patients who have complex medical needs who may have difficulty navigating a new diagnosis and its implications. Through education, I hope to empower patients and their families.

Dr. Grant: I am fascinated by the brain, and I love to have such an impact on the lives of children to do whatever it takes to cure their seizures but also to maximize their quality of life. I just want the kid to be a kid again.

Dr. Isom: I was motivated to choose a career in nephrology because my medical school faculty were so inspiring in their outlining of the interconnectedness of form and function, as relates to the kidney. The field of nephrology has allowed me to develop professionally and to have a positive impact on patients' lives.

Perla: I have been a Case Manager for the past 10 years and was an ER nurse prior to taking on this role. I completed my masters to be a nurse educator and on my path to finding a career to transition to, a case management position crossed my path. I chose this position/career because I felt challenged by the ambulatory role itself and all the evolving neurology-related matters that it encompasses from medications down to finding community resources.

Dr. Teng: I have always interested in medicine since I was young. Dermatology is an ideal field for me as it demands the quintessential combination of visual acuteness with wealth of medical and surgical knowledge.

Bridget, you are the Step Forward to Cure TSC Chair for Northern California and the TSC Clinic Ambassador at Lucille Packard Children's Hospital, Stanford, CA. You've also come to Washington, DC to advocate for Federal funding for TSC research. What motivates you to volunteer for the TS Alliance?

Raising money to fund critical research to find a cure for TSC has been a huge motivator. Also, being able to bring fellow TSC families together to connect, share stories, and be a part of the TSC family.

Dr. Porter, some consider the ideal clinic model to be where a person with a multi-system disease such as TSC sees more than one specialist on the same day. At Stanford, you care for children with TSC and Dr. Le cares for adults at a different location on the Stanford University campus. When you see a child for the first time, are other specialists at the clinic or are arrangements made for the child to see them at another time?

My first visit is sometimes a solo visit to go over TSC and discuss all the aspects of the disease. Having a new baby diagnosed with TSC is super scary, and I think going slow and introducing all the aspects of the disease and often focusing on the most important disease area at that moment can lessen the overwhelming feeling parents often have. Everything in California and Nevada is so spread out and some of our patients often drive many hours or even fly in, so I try to coordinate multiple appointments for them, though a two-day visit is often needed to see multiple specialists. Our biggest problem is lack of clinic space. We are not allowed to build new larger clinic buildings, which is making us be creative with space, I now see patients into the evening so I can fit all my patients into the rooms I am allowed to use. I am blessed though to have many experts in their disease area to help take care of them.

Dr. Teng, when you see an adult for the first time, are other specialists at the clinic or are arrangements made for the adult to see them at another time?

Since I am board certified in both pediatric and general dermatology, I see both adults and children with genetic diseases in my clinic. My clinic is located in a multispecialty clinic, so it is very convenient for me to consult other medical and surgical specialists such as ophthalmology, ENT, neurology, oncology etc. Dr. Porter and I frequently share patients on the same day.

In June 2012, you and six dermatologists and two dentists met in Washington, DC to update the diagnostic criteria, surveillance and management of TSC features. What advances in the diagnosis and management of TSC skin manifestations have been made since then?

We have expanded our working group to eight people this year to include two international dermatology specialists. We have performed extensive literature searches and conducted multiple survey studies in regard to update on cutaneous manifestation, surveillance and management recommendations. Randomized clinical trial data from US and Japan on the use of topical sirolimus was included. We have presented our findings and recommendations at last summer's 2018 World TSC Conference in Dallas. The written consensus in diagnosis, surveillance and management from the working group has been submitted to TS Alliance.

What are the most challenging aspects of your work as it relates to those affected by TSC?

Dr. Porter: There are so many, but I absolutely hate it when someone was not diagnosed correctly. It still happens way too often! They may see a pediatric specialist who misses all the classic signs of TSC. I also feel there is a window of time I can have the biggest impact on brain development. If that has passed without me getting to try a variety of things to improve neuronal outcome, it drives me crazy.

Dr. Le: Though amazing advances have occurred in the understanding and treatment of TSC, ineffective or only partially effective therapies for epilepsy is frustrating. Limited resources for neuropsychiatric treatments for patients are also challenging.

Elli: The genetics of TSC can be confusing and overwhelming, especially when a diagnosis is new or unexpected. These can be difficult conversations that include topics like: recurrence risks for other children, the meaning of an uncertain or negative genetic test, and the guilt a parent may feel when discussing a genetic result.

Dr. Grant: There are so many possible areas of seizure onset in a child with TSC, so often it is very difficult to determine which tuber or tubers are the best target. We now can navigate to many of these potential targets all at once with our stereo EEG approach to determine which target is the hottest. The high precision stereo EEG approach is a minimally invasive approach to sample the 3D network on both sides of the brain all at once.

Dr. Isom: Remembering all patients are unique, especially when it comes to assessing the neurologic impairment(s) they may suffer as a consequence of their disease. This requires that I tailor my approach on how I communicate my findings and recommendations to patients and their families.

Perla: The TSC neuropsychological-related issues associated with the condition is the most challenging part. As a case manager, coordinating or even finding psychiatry effectively and expediently to help co-manage the symptoms with the team is also a challenge, particularly with those who live out of area with very little to no resources available locally.

Dr. Teng: Delayed diagnosis due to lack of recognition of cutaneous manifestations of TSC. Access to specialists for adequate care and management.

Bridget, tell us the most challenging aspects of being a TS Alliance volunteer.

One of the most challenging aspects of being a TS Alliance volunteer is telling a parent the same words I had to hear, "there are no guarantees in how TSC will affect your child, and you have to take the disease one day at a time."

What are the most fulfilling aspects of your work?

Dr. Porter: Bonding with families – I love watching my patients grow up. My patients always astound me with their insights and are always teaching me new things. My favorite is when one turns to his or her parents and says, "This is my life and I am deciding what we are going to do about my seizures." I know I did my job when patients take control of their epilepsy or their TSC.

Dr. Le: What I enjoy most about my work are the long-term relationships with patients and their families. I also derive fulfillment from educating the next generation of medical trainees.

Elli: I am so fortunate to work with families who are compassionate, resilient, and strong advocates for their children.

Dr. Grant: Giving a family hope that we will give our all to cure their child's seizures and watch the brain development soar! I treat every kid like my own.

Dr. Isom: With regard to management of TSC patients, definitely having at our disposal a form of therapy shown to favorably impact the course of disease.

Perla: I truly find joy in my role knowing I accomplish something daily – whether simple or complex, whenever things go the right way for the best interest of the child – makes a difference to every child/family. The positive outcome, the gratitude from the parents, and the exuberance from the provider/team are priceless.

Dr. Teng: Providing education and new treatment options to patients with TSC has made tremendous impact on the care of patients with TSC. I do not see patients with terrible cutaneous manifestations as I did a decade ago. Notable improvement of TSC-related skin manifestations always has a really positive effect on patients as well as their families.

Bridget, what are the most fulfilling aspects of being a TS Alliance volunteer?

The most fulfilling aspect of volunteering is being able to help fellow TSC parents know they are not alone in the journey.

This is a "what if" question: What if you had the power to make a change that would impact the lives of those affected by TSC, what would you do?

Dr. Porter: I would develop a gene therapy for TSC. Gene therapy for genetic disorders has become a reality. While neurologic diseases with diffuse multi-organ involvements will be slower to be widely used, it for sure is going to happen; it's just a matter of when. There are multiple gene therapy treatments for genetic disorders under investigation, and several will soon be in clinical trials.

Dr. Le: I would find a preventative cure for TSC.

Elli: I would create more widespread familiarity with the signs and symptoms of TSC in hopes that diagnoses will not be missed or delayed. Early diagnosis and treatment will be important as new therapies enter the market.

Dr. Grant: The power to treat a child's seizures in a way that is painless and risk-free.

Dr. Isom: Develop alternative medical/pharmacologic approaches to treatment that have fewer side effects than our current approach.

Perla: (Wishful thinking) would like to have a whole TSC model access to care and access to variety of options down to county level accessibility for community resources, regardless of insurance coverage or location.

Dr. Teng: Provide them access to the care they deserve.

Bridget: My goal would be to improve access to TSC specialists and other healthcare providers for families living in rural areas. So many families have to travel great lengths to go to TSC clinics or don't have access tp behavioral therapists, etc., because options are limited or nonexistent. I would want to help find a solution to change that so our TSC families can get the best healthcare possible close to where they live.

How do you balance your time between your administrative, academic and/or clinical responsibilities and personal time; and what do you enjoy doing outside of work?

Dr. Porter: I love living in California. We moved here six years ago, and it is a great place to live if you love nature. The weather allows for year-round outdoor activities. My husband and I, and if we can convince one of our kids, will go for walks in the park next to or house or a hike in one of the nearby state parks. We are into bird watching, and the beach or mountains are only a couple of hours away. We just hired or ninth pediatric epilepsy doctor at Stanford, which should give me more free weekends and more time to garden and enjoy the outdoors.

Dr. Le: I'm a Clinician Educator: I mainly see patients and teach trainees. Outside of work, I spend time with my family.

Elli: I am still learning how to better balance clinical and personal time! I love to read, bake, hike, and dance.

Dr. Grant: I am blessed with the privilege of doing what I love and always striving to do my best for others. I am proud of our team members, who all contribute to the successful outcomes of the children we treat. I enjoy being with my own family and love the beach.

Dr. Isom: I do my best to complete my day's work while I am still in the office or at the hospital, so as not to "bring it home" with me. This is a real challenge, and I realize it will never be 100 percent attainable. Outside work, I enjoy staying current with family and old friends, as well as hiking and swimming for exercise.

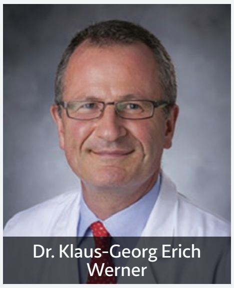
Perla: After a work week, I find enjoyment to stay at home, enjoying the company of our eight four-legged children (three dogs, two cats and three foster kittens) or having dinner and a movie night with my husband. In addition, whenever time permits, I do enjoy hiking and backpacking (my goal is to make it to the summit of Mount Whitney next year).

Dr. Teng: There are never enough hours in a day! I enjoy spending time with my family – cooking, walking, traveling.

Bridget: I balance my time between volunteer work and my family by taking advantage of pockets of time. I send emails while sitting on the foot of the bed waiting for my daughter to fall asleep. I participate in conference calls after making sure my kids got to the park first, so they are ready to play quietly while I am listening in. My family is also a huge help watching my children if I need blocks of time to do something. For relaxation, I like to visit my friends, get a pedicure, or watch TV at the end of a long day.

A Learning Experience for Pediatric Residents and Families in South Carolina:

WALKING IN A MINEFIELD CALLED TUBEROUS SCLEROSIS COMPLEX


Dr. Klaus-Georg Erich Werner

This past summer, Klaus-Georg Erich Werner, MD, PhD, MRCP(I), MRCPCH, Director of the TSC Clinic at Duke University in Durham, NC contacted Jo Anne Nakagawa, TS Alliance Director of Clinical Projects and TSC Clinic Liaison, to propose two educational meetings in South Carolina near TS Alliance-recognized TSC Clinics.

On Friday, December 7, he gave a presentation to more than 20 pediatric residents and other medical professionals at the Palmetto Health Department of Pediatrics Grand Rounds Conference in Columbia. One attendee was Diane Biederman, a genetic counseling student at the University of South Carolina whose master's thesis is on TSC. She wrote, "Only a few residents were familiar with TSC before the lecture. I think his presentation was a valuable training experience for them."

Despite extreme weather conditions the following day, 19 people attended the TSC Family Educational Meeting in nearby Lexington, where Dr. Werner was joined by two other clinicians experienced in caring for individuals with TSC. Dr. Monica Cramer, a pediatric kidney specialist formerly affiliated with the TSC Clinic at the University of Alabama Birmingham, is now at the Palmetto University of South Carolina Medical Group. In addition, Dr. Katherine Van Poppel, a pediatric neurologist and epileptologist, formerly Co-Director of the TSC Clinic at Le Bonheur Children's Hospital in Memphis, is currently Director of the TSC Clinic at Levine Children's Hospital, Charlotte, NC. The audience expressed appreciation for gaining valuable information about TSC with comments such as, "Dr. Werner is a dynamo," and "The Family Educational Meeting was excellent!"

The TS Alliance commends Dr. Werner for his initiative to educate healthcare professionals in South Carolina about TSC and the possibility of him starting a satellite TSC Clinic there for children and adults.

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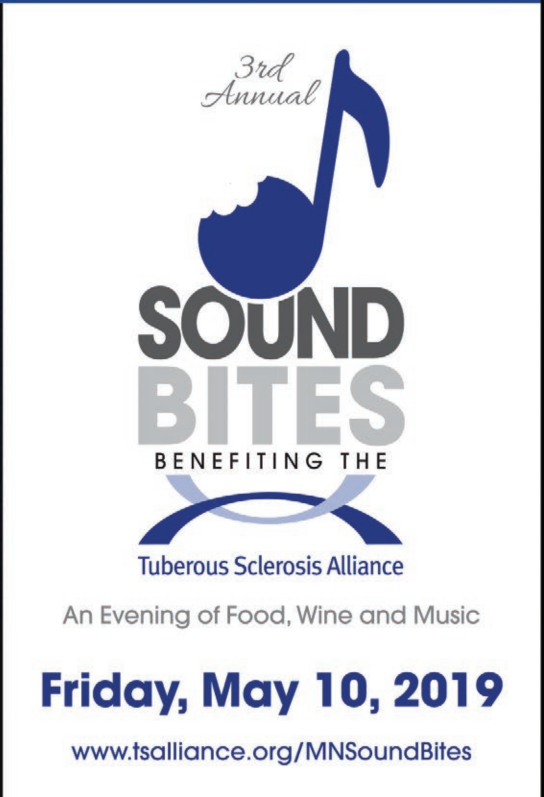

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