

2022 World TSC Conference Honorees

Dr. Michael and Janie Frost | TSC Champion Award

Michael and Janie Frost have both devoted their careers and lives to helping individuals and their families affected by TSC.

Dr. Frost is a board-certified child neurologist and epileptologist. He was a founding team member of Minnesota Epilepsy Group in 1991 and established the Upper Midwest TSC Clinic Without Walls after meeting Dr. John Hulbert at a TSC Alliance research conference in 2002.

During his tenure as Co-Director of the TSC Clinic, he served as an investigator on several pivotal clinical trials including Afinitor, which led to its approval for treating SEGAs in 2010; Epidiolex that led to its approval in 2020; and the current PREVeNT Trial, the first preventative clinical trial for epilepsy in the United States. In 2006, Minnesota Epilepsy Group was one of two pilot sites for the TSC Natural History Database. Additionally, Dr. Frost served on the Clinical Consensus Group in 1998 to help establish the original TSC Clinical Consensus Guidelines and continued this role through two additional revisions with a special expertise in comprehensive and coordinated care. Dr. Frost continues to provide his guidance as a member of the TSC Alliance Professional Advisory Board.



Janie Frost is a trained epilepsy nurse. She served as a member of the TSC Alliance Board of Directors from 2006-2011 and as Secretary in 2009. With her expertise in clinical care, she helped organize the 1999 (Washington, DC) and 2001 (San Diego) National Family Conferences and chaired the National and World TSC Conferences in 2006 (Chicago), 2014 (Washington, DC) and 2018 (Dallas). These conferences are among the most prolific gatherings of the global TSC community and were successful because of her outstanding leadership and dedication. Janie also generously served on the planning committee for the Minnesota-based Sound Bites events as well as the 35th, 40th and 45th TSC Alliance anniversary galas.

The impact of Dr. Michael and Janie Frost on the TSC community has been profound. Their deep compassion and extraordinary commitment helped create an improved quality of life for tens of thousands of individuals and families. Join us in celebrating them and their many accomplishments during Saturday's Blue Night Dinner.



Welcome to the 2022 World TSC Conference

Dear Conference Participants,

The TSC Alliance® and Tuberous Sclerosis Complex International (TSCi), along with Title Sponsor Jazz Pharmaceuticals and Presenting Sponsor Nobelpharma, welcome you to Dallas for the 2022 World TSC Conference! After a very challenging few years, we are thrilled you decided to join us for another amazing educational event, featuring more than 100 speakers from around the world. Of course, this conference also offers you the unique opportunity to make life-long friendships and connections with other families and individuals affected by tuberous sclerosis complex (TSC).

The 2022 World TSC Conference features diverse sessions daily, including general sessions and keynote speakers to address TSC through the lifespan, as well as question-and-answer sessions at the end of the day on Friday and Saturday. The conference agenda is organized into five learning paths to align with the different ways people learn:

- Large Group Lectures (livestreamed)
- Panel Presentations (livestreamed)
- Specialty Topic Lectures (livestreamed)
- Workshops (non-recorded)
- Discussion Groups (non-recorded)

Unless specifically noted, each session is designed for our global TSC community. Four sessions are marked to notify attendees that all are welcome; however, these sessions may be most relevant to individuals living in the United States. You can find those marked with an asterisk (*) on the agenda.

We encourage you to take advantage of the evening social activities and get to know others who may have a similar TSC story in a comfortable social environment. We have gatherings for TSC moms, dads, adults and siblings; a Spanish gathering; a multicultural meet and greet event; and a Teen Lounge open several hours each day. We also enhanced support options for our underserved dependent adult families with evening gatherings in our Sensory room, located at the rear of Stemmons Ballroom.

Throughout the conference we encourage you to visit our Exhibit Hall & TSC Resource Center to learn more about services, programs, products and information from a wide range of vendors. The TSC Alliance Store will offer commemorative 2022 World TSC Conference and TSC Alliance merchandise. And for the first time we are adding a Research Room to highlight clinical studies and clinical trials.

Thank you for attending this life-changing event! Now more than ever and because of each of you, the global TSC community remains hopeful, relentless and strong.

Sincerely,

Peter Crino, MD, PhD Chair, TSC Alliance Board of Directors

Peter B. Criso

Kari Luther Rosbeck President & CEO Katie Smith TSCi Working Group Chair Mary Roberds Conference Chair

Get the 2022 World TSC Conference App

Be sure to install the 2022 World TSC Conference App to access the conference agenda, speaker information, sponsor and exhibitor lists and much more. You can even message other attendees and create personalized schedules.

1 Install the "Cvent Events" app.

On your device, access the App Store or Google Play Store and search for the "Cvent Events" app. Once you've found the app, install it.

2 Search for the "2022 World TSC Conference" event.

Return to your device's home screen and open the "Cvent Events" app. Next, search for "2022 World TSC Conference."

3 Download the event.

4 Enter your info.

If you aren't automatically prompted to log in, tap the Profile tab at the bottom of the screen, then Log in. Enter your first name, last name and email. Then tap the arrow.

5 Verify your account.

You'll either receive an email and a text message containing a 6-digit verification code or just an email (be sure to check your Junk Folder if you don't see the email). Read it, then return to the app, enter the code and tap the arrow. After entering the verification code, you'll be logged in.

NOTE: Verification codes can only be used once and expire after 24 hours. If you're logging in on multiple devices, you'll receive a new verification code for each.

6 Complete your profile.

Tap the Profile tab again, then tap View Profile. To make edits, including adding your name and photo, tap the Pencil icon on the upper right and complete the information. Next, choose your Visibility Settings. By default, your Profile is set to Hidden. If you want others using the app to see your information, check the Visible icon. Then tap Save on the upper right.

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Contents

Welcome	••••
Conference App Instructions	
Conference at a Glance	4
Conference Sponsors	
General Conference Information	8
Day Camp Information and Schedules	1
About the TSC Alliance	12
TSC Alliance Leadership and Staff	12
TSC Alliance Volunteer Leaders	1
Global Alliances	20
About Tuberous Sclerosis Complex International (TSCi)	20
TSC Clinics	23
Session Descriptions	2
Full Conference Agenda	. 30
Conference Speakers	. 38
Exhibit Hall and TSC Resource Center	70
Glossary	74
Conference Acknowledgements	. 84



Conference at a Glance

DATE and TIME	General Session Grand Ballroom D/E and various locations	Panel Presentations Grand Ballroom A	Specialty Topic Lectures Grand Ballroom B	Large Group Lectures Grand Ballroom D/E	Workshops (non-recorded) Plum Blossom	Discussion Groups (non-recorded) Edelweiss
Thursday July 28						
12 - 5 p.m.	Biosample Repository Blood Draw, Inverness					
12 - 6 p.m.	General Conference Registration and Day Camp Registration, Conference Registration Room and Atrium II					
3 - 5 p.m	Meet and Greet Opportunities					
4 - 5 p.m.						TSC 101 for Newly Diagnosed Individual
6:30 - 8:30 p.m.	Conference Welcome and Opening Dinner, Imperial Ballroom					or Families
8:30 p.m.	After Dinner Activities: Adults with TSC, International Social, TSC Dads, Parents and Dependent Adults, and Teen Lounge					
Friday, July 29						
7 - 8:30 a.m.	Breakfast, Chantilly Ballroom					
7 - 8:30 a.m.	Biosample Repository Blood Draw, Inverness					
7 - 8:30 a.m.	Research Room Open, Fleur de Lis A & B					
7:30 a.m 5:30 p.m.	Day Camp, Atrium I- Check-In: Stemmons Ballroom					
8 a.m 12 p.m.	General Conference Registration - Conference Registration Room and Atrium II					
8 a.m 5 p.m.	Exhibit Hall and TSC Resource Center, Grand Ballroom C					
8 a.m 5 p.m.	TSC Store Open, outside Grand Ballrooms					
8:30 - 10 a.m.	OPENING GENERAL SESSION: Global Collaborations (TANDem) and Consensus Guidelines Updated					
10 - 10:30 a.m.	BREAK					
10 a.m 3 p.m.	Biosample Repository Blood Draw, Inverness					
10 a.m 3 p.m.	Research Room Open, Fleur de Lis A & B					
10:30 a.m 12 p.m.		* Navigating the Complexities for Special Needs Care in TSC (SSI, Waivers, Medicaid, Access, Financial Planning & Guardianship)	Using Technology to Your Advantage: Seizure Monitoring, Empowering Communication for the Nonverbal, and Futuristic Medical Care	Seizure Types and Treatments	Organizing Your Thoughts and Emotions and Managing Relationships (TSC individual)	Preparing Your Family for Epilepsy Surgical Evaluation and Surgery

DATE and TIME	General Session Grand Ballroom D/E and various locations	Panel Presentations Grand Ballroom A	Specialty Topic Lectures Grand Ballroom B	Large Group Lectures Grand Ballroom D/E	Workshops (non-recorded) Plum Blossom	Discussion Groups (non-recorded) Edelweiss
12 - 1 p.m.	Box Lunches, Ballroom Foyer	Lunch and Learn (Box Lunches): Caregiver Mental Health and Wellbeing			Lunch and Learn (Box Lunches): TSC 101 Part II - A Caregiver Guide to Advocate for Your Loved One	
12 - 5 p.m.	Teen Lounge Open, Obelisk A & B					
1 - 2:30 p.m.		Surgical Approaches and Assessments for Epilepsy and SEGA	Accessing Managed and Palliative Care Throughout Your TSC Journey	Skin and Dental Manifestations and Treatment Options	Preparing for Transitions - Childhood to Adulthood (Medical Care)	Women's Health Issues for TSC Adults
2:30 - 3:00 p.m.	BREAK					
3 - 4:30 p.m.		Accessing Early Childhood Services & Non-medical Therapies	LAM, Treatment Options and Clinical Trials	Genetics Made Simple	TAND - Aggressive Behavior Management	Men's Health Issues for TSC Adults
4:30 - 4:45 p.m.	BREAK					
4:45 - 5:30 p.m.	General Questions and Answers- Panel of Presenters, Grand Ballroom D & E					
5:30 - 7 p.m.					Encuentro de educación en Español y conocer y saludar - Spanish Education Meeting and Gathering	
6:30 - 8:00 p.m.	Dinner, Chantilly Ballroom					
8:30 p.m.	After Dinner Activities: Adults with TSC, Multicultural Social, Parents and Dependent Adults, and Teen Lounge					
8:45 p.m.	Movie Night, Jade Oval Lawn					
Saturday, July 30						
7 - 8:30 a.m.	Breakfast, Chantilly Ballroom					
7 - 8:30 a.m.	Biosample Repository Blood Draw, Inverness					
7 - 8:30 a.m.	Research Room Open, Fleur de Lis A & B					
7:30 a.m 5:30 p.m.	Day Camp, Atrium I- Check-In: Stemmons Ballroom					
8 - 10 a.m.	General Conference Registration - Conference Registration Room and Atrium II					
8 a.m 5 p.m.	Exhibit Hall and TSC Resource Center, Grand Ballroom C					
8 a.m 5 p.m.	TSC Store Open, outside Grand Ballrooms					
8:30 - 10 a.m.	GENERAL SESSION: TSC Community Driven Research Network (TSC Alliance Research Model)					

Conference at a Glance (cont.)

DATE and TIME	General Session Grand Ballroom D/E and various locations	Panel Presentations Grand Ballroom A	Specialty Topic Lectures Grand Ballroom B	Large Group Lectures Grand Ballroom D/E	Workshops (non-recorded) Plum Blossom	Discussion Groups (non-recorded) Edelweiss
Saturday (cont.)						
10 - 10:30 a.m.	BREAK					
10 a.m 3 p.m.	Biosample Repository Blood Draw, Inverness					
10 a.m 3 p.m.	Research Room Open, Fleur de Lis A & B					
10:30 a.m 12 p.m.	Sibling Workshop, Cooper	TAND - ADHD, Anxiety Autism (Research)	, Caregiver Resources - Self & Respite Care	Kidney Tumors, Cysts, and Treatment Options	Seizure Action Plans & Rescue Meds	* Navigating Educatio System for a High School Certificate
12 - 1 p.m.	Box Lunches, Ballroom Foyer	Lunch and Learn (Box Lunches): Peer-to-Peer Support Programs	Lunch and Learn (Box Lunches): Government Advocacy		Lunch and Learn (Box Lunches): Special Events & Fundraising	
12 - 5 p.m.	Teen Lounge Open, Obelisk A & B					
1 - 2:30 p.m.		Genetics, Reproductive Issues and Impact on Relationships	Refractory Epilepsy - Now What?	Access to TSC Clinical Care	Advocacy in the Education System	TAND - Mental Health Issues in Living with TSC as an Independer & Semi-Independent Adult
2:30 - 3 p.m.	BREAK					
3 - 4:30 p.m.		* Finding your Way Home – Creative Housing Options/ Microboards	Managing Progressive Kidney Disease	Gene Therapy - Hopes and Hurdles	TAND - Feeding, Elopement & Potty Training	What's Next? Tools to Empower You on Your Medical Journey to an Adult Neurologist
4:30 - 4:45 p.m.	BREAK					
4:45 - 5:30 p.m.	General Questions and Answers- Panel of Presenters, Grand Ballroom D & E					
6:30 - 8 p.m.	Blue Night Dinner, Chantilly Ballroom					
8:30 p.m.	After Dinner Activities: Adults with TSC, TSC Moms, Parents and Dependent Adults, and Teen Lounge					
Sunday, July 31						
7 - 8:30 a.m.	Breakfast, Chantilly Ballroom					
7:30 a.m 12:30 p.m	Day Camp, Atrium I- Check-In: Stemmons Ballroom					
8 a.m 12 p.m.	TSC Store Open, in Registration Area					
8:30 a.m 10 a.m.		Other Organ Manifestations - Liver, Pancreas, Heart and Eyes	Transition into Adulthood - Employment and Higher Education	TAND - Behavioral Interventions and Overview of Medications	* Navigating Health Care System to Better Access Medication	Sexual Development for Dependent & Semi-Independent TSC Adults
10 - 10:30 a.m.	BREAK					
10:30 a.m 12 p.m.	CLOSING SESSION: Driving the Future of TSC - Clinical Trials & Clinical Care					

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The TSC Alliance and TSCi extend our deepest appreciation and gratitude to the following sponsors and donors for their support of the 2022 World TSC Conference. Their generosity makes this experience possible.

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University of Iowa Stead Family Hospital

Additional Support

Lundbeck



General Conference Information

Conference, Day Camp and Registration Hours*

Thursday, July 28

Noon to 6 p.m. Conference and Day Camp Registration/Check-In at Conference Registration Room and Atrium 2

Friday, July 29

7:30 a.m. to 5:30 p.m. Day Camp, Check-In/Out at Stemmons Ballroom

8 a.m. to Noon Daily Conference Registration at Conference Registration Room and Atrium 2

Saturday, July 30

7:30 a.m. to 5:30 p.m. Day Camp Check-In/Out at Stemmons Ballroom

8 to 10 a.m. Daily Conference Registration at Conference Registration Room and Atrium 2

Sunday, July 31

7:30 a.m. to 12:30 p.m. Day Camp Check-In/Out at Stemmons Ballroom

*Please note: The Registration Desk will always have someone available to assist you with questions and/or directions.

Research Room

TSC Research Studies and Clinical Trials: The Research Room, located in Fleur de Lis A/B on the mezzanine level, provides attendees with a place to learn about ongoing TSC research studies and clinical trials including opportunities for how to participate. There is no obligation to participate. We encourage all attendees to visit! The Research Room will be open as follows:

Friday, July 29: 7 to 8:30 a.m., 10 a.m. to 3 p.m. **Saturday, July 30:** 7 to 8:30 a.m., 10 a.m. to 3 p.m.

On-Site Blood Collections: The TSC Alliance also offers on-site blood collections, located in the Inverness Room right next to the Research Room on the mezzanine level, for the TSC Biosample Repository and Natural History Database project. Blood draws are available for individuals who have a definite diagnosis of TSC and/or LAM. If you are not currently participating in the TSC Biosample Repository and Natural History Database project, the TSC Alliance research staff may be able to complete the consent process on-site at the conference depending on availability. A total of 17 hours will be available for attendees to complete their blood draws.

Thursday, July 28: Noon to 5 p.m.

Friday, July 29: 7 to 8:30 a.m., 10 a.m. to 3 p.m. **Saturday, July 30:** 7 to 8:30 a.m., 10 a.m. to 3 p.m.

Skin Photography: The TSC Alliance is also collecting photographs of TSC skin manifestations across individuals of differing skin tones, including Black and Asian individuals with TSC, to create a reference resource to share with clinicians with the goal of education and raising awareness of these differences. To date, medical photographs of such manifestations are commonly shown as examples from White or other individuals with lighter skin tones, creating a barrier for clinicians to accurately compare in Black or other individuals with darker skin tones.

Interested in providing photographs or being photographed for this initiative? Please visit the Research Room, located in Fleur de Lis A/B on the mezzanine level, to participate! We will have someone present to take photographs. All participants will be required to sign a photo release for use of photos.

Friday, July 29: 8 to 8:30 a.m., 10 a.m. to 11 a.m., 12:30 to 1 p.m.

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Teen Lounge

The Teen Lounge, located in Obelisk A&B, is a space reserved for connecting teens with TSC (ages 12-23). They can hang out in a one-stop hub with televisions, video and other games, crafts and special programming. *Parents please note: This room is not supervised.*

Thursday, July 28: Friday, July 29: Saturday, July 30:

8:30 to 11 p.m. 12:30 to 5 p.m., 8:30 to 11 p.m. 12:30 to 5 p.m., 9 to 11 p.m.

Blue Night Dinner

Be sure to wear blue for Saturday night's Blue Night Dinner, where we'll honor Dr. Michael and Janie Frost with a TSC Champion Award for their lifelong commitment to caring for the TSC community. You'll also see and hear a special performance by TSC mom and American Idol sensation Althea Grace, followed by a live DJ for our first World TSC Conference Prom.

Emergencies/First Aid

A first aid kit will be available at the Registration Desk and in each Day Camp room. A Texas-licensed physician will be available for immediate assistance; please call (945) 210-0546 if you need to reach the on-call physician. Emergency paramedic assistance will be alerted if emergency care and transport are needed.

Nearby Medical/Emergency Resources

The following facilities are within 2.5 miles of the Hilton Anatole:

UT Southwestern Medical Center / Zale Lipshy Hospital

General Medical Treatment 5151 Harry Hines Blvd. Dallas, TX 75390 www.utswmed.org (214) 645-5555

Parkland Memorial Hospital

Major Trauma Center 5200 Harry Hines Blvd. Dallas, TX 75235 www.parklandhospital.com (214) 590-8000

Concentra Urgent Care – Stemmons

Urgent Care 2920 N. Stemmons Fwy. Dallas, TX 75247 www.contentra.com (214) 630-2331

Walgreens Pharmacy

Open 24 Hours 3802 Cedar Springs Rd. Dallas, TX 75219 www.walgreens.com (214) 443-5160

Lost and Found

Lost items that are found and turned in may be claimed at the Registration Desk or the TSC Alliance booth in the Exhibit Hall.

Recommended Dress

Attendees should wear casual clothing. Please remember meeting rooms are air-conditioned and may be cool. Sweaters or jackets may be needed. Check out the TSC Alliance Store if you forgot yours! Also, remember to wear your blue for Saturday evening's Blue Night Dinner.

Meeting Room WiFi Access

Free WiFi is available in all meeting rooms, courtesy of Drs. Bonnie and Jonathan Rothberg & Family, Visionary Technology Sponsor. Look for SSID: "World TSC Conference" and use Password: "welcome22".

TSC Store

TSC Alliance and conference merchandise is available for purchase at the TSC Store, located outside the Grand Ballrooms except Sunday as noted below.

Thursday, July 28: Noon to 6 p.m. Friday, July 29: 8 a.m. to 5 p.m. Saturday, July 30: 8 a.m. to 5 p.m.

Sunday, July 31: 8 a.m. to Noon, in Conference Registration Area

Conference Educational Opportunities

General Q&A Sessions with Panel of Presenters, Grand Ballroom D & E

The 2022 World Conference will include two general Question and Answer Sessions. These sessions will feature TSC expert physicians who will be available for general discussion and questions on any topics related to TSC. These sessions will also be livestreamed for virtual attendees.

Friday, July 29: 4:45 to 5:30 p.m.

Moderator: Kari Luther Rosbeck

Panelists: Nathan Call, PhD, BCBA-D; Peter Crino, MD, PhD; Dan Curry, MD; Tanjala Gipson, MD; Nishant Gupta, MD; Chris Kingswood, MD; Hope Northrup, MD; Joshua Samuels, MD, MPH; Joyce Teng, MD, PhD; Howard Weiner, MD; James Wheless, MD

Saturday, July 30: 4:45 to 5:30 p.m.

Moderator: Steve Roberds, PhD

Panelists: Derek Bauer, MD; John Bissler, MD; Petrus de Vries, MBChB, FRCPsych, PhD; David Dunn, MD; Gerald Grant, MD; Lisa Henske, MD; Shafali Jeste, MD; Darcy Krueger, MD, PhD; Angela Peron, MD; Elizabeth Thiele, MD, PhD

Large Group Lectures (livestreamed) will feature 3-4 speakers with a moderator and include formal presentations on topics that impact individuals across the lifespan with some time at the end for Q&A.

Panel Presentations (livestreamed) will feature 3-4 speakers with a moderator who will ask questions to engage interactive conversation amongst the panel and the attendees.

Specialty Topic Lectures (livestreamed) will feature at least 2+ speakers with a moderator to focus on specialty topics that affect the TSC community.

Workshops (non-recorded) are interactive working groups featuring 3-4 speakers, a moderator and a key take away learning objective with supporting materials.

Discussion Groups (non-recorded) will feature 2-3 speakers and a moderator to discuss more personal topics that affect the TSC community in a closed setting.

Group Sessions are specialty topics that impact a subset of constituents and may be more personal in nature and therefore will not be recorded. These sessions will run concurrently with the tracks.

Disclaimers

The information presented during all sessions is intended for educational purposes only and is not endorsed by the TSC Alliance. Due to unforeseen circumstances, the conference schedule and speakers may change.

Some conference sessions are being recorded, and attendees may be recorded or photographed. All conference participants agree to be recorded or photographed without compensation. Photographs and videos may be used in future publications and promotional materials.

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Day Camp Hours, Information and Schedule

Hours

Friday, July 29: 7:30 a.m. to 5:30 p.m. **Saturday, July 30:** 7:30 a.m. to 5:30 p.m. **Sunday, July 31:** 7:30 a.m. to 12:30 p.m.

Day Camp Check-In/Check-Out: Stemmons Ball Room

Day Camp participants will enjoy planned activities and meals, with each program designed to provide children and dependent adults with a fun, stimulating environment while allowing for flexibility in the schedule based on each camper's individual needs. All Day Camps are located in the Stemmons Ball Room. The 2022 World TSC Conference Day Camps are managed and operated by Corporate Kids Events (CKE).

Day Camp participants are divided into three groups:

1. Camp Discovery – This camp is for children with TSC who are functioning at an age-appropriate level as well as unaffected siblings of a TSC child. Children with TSC will be considered for Camp Discovery: if they are on IEPs with minimal support, can perform all activities of daily living independently with only occasional verbal reminders and can functionally communicate wants and needs. A family interview with a Camp Leadership Team member may be warranted if concerns exist about which Camp would best suit a child.

Staffing ratios for Camp Discovery are as follows:

- Infants 0 to 2 Years: 1:2Children 3 to 4 Years: 1:4Children 5 to 7 Years: 1:5Children 8 to 12 Years: 1:6
- Children 8 to 12 Years: 1:6
 Children 13 to 17 Years: 1:8
- Children in Camp Discovery are divided by chronological age.
- 2. Camp Explorer This is the special needs camp for TSC-affected children with moderate to significant needs. Children who require assistance with activities of daily living, communication, mobility, behavior management or seizure control will all be best served through Camp Explorer. This camp will be staffed at a 2:1 camper to staff ratio.
- **3. Dependent Adult Camp** This is a camp for dependent adults with TSC with moderate to significant needs. Activities will be individually established based on the camper's interests and will include as much as the camper is able to try. Special events happening in the other camps may be available to the adult campers on an event-by-event basis. This camp will be staffed at a 2:1 camper to staff ratio.

Please note: Each Day Camp ends promptly at 5:30 p.m. Friday and Saturday and at 12:30 p.m. Sunday.

Because safety is of primary concern to the TSC Alliance and CKE, each Day Camper will be required to wear an I.D. bracelet, be signed in and out by a parent or authorized caregiver, and a digital photo security system will be utilized (photos with camper and parents/guardians). A parent is required to give medication.

Camp Discovery Schedule*

*Camp Explorer and the Adult Day Camp will participate in the following activities according to interest and ability.

	Friday, July 29 7:30 am-5:30 pm	Saturday, July 30 7:30 am-5:30 pm	Sunday, July 31 7:30 am-12:30 pm
Kids Camp (Ages 3-5 years)	 Pirate Party Pirate Costumes Face Painting Fun Special Guest Blimey Photos and Frames Shipwreck Crafts Lunch Time Story Time Walk the Plank Tiny Treasure Boxes Special Presentation Tasty Treasure Hunt 	 Beach Party Bonanza Sign-In / Digital Photos Funky Flower Leis Sunny Visors & Silly Sunglasses Balloon Volleyball Limbo Madness Yummy Lunch Fun Fire Trucks Movie Time Sand Creations Art Extravaganza 	 After While Crocodile Awesome Awards Play it Again Magic Show Awesome Address Books Closing Address
Tween Camp (Ages 6-11 years)	Toom Floor / Toom Hoto L & Cool Conco		 Later Gator Business Card Exchange Magic Show Tween Support Group Awards Closing Address
Teen Camp (Ages 12-17 years)	 The Teen Scene Sign-In/Digital Photos Parent Release Ice Breakers Teen Trivia Contest Special Guest Teen Trust Falls Jazzy Jewelry Design Lunch Time Funny Photos and Frames Cruising the Resort Special Presentation Fabulous Facemolds 	 Lean, Mean and Teen! Sign-In/Breakfast Funny Human Tricks Super Scavenger Hunt Lunch Time Memory Books T-Shirt Graffiti Party Minute to Win It! Art Extravaganza Music Mania 	 Until Next Year Email Blast Photo Exchange Magic Show Teen Support Counselor Recruits Group Hug

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About the TSC Alliance®

The TSC Alliance® is an internationally recognized nonprofit that does everything it takes to improve the lives of people with tuberous sclerosis complex (TSC).

We are a source of hope and connection for all affected by TSC. We drive research, increase care quality and access and advocate with and for people affected by the disease. Through our collaboration and partnerships, we've advanced FDA-approved treatments and created support systems around the world so that no one has to navigate TSC alone.

The TSC community is our strongest ally. With the power of families and the support of donors, volunteers, researchers, educators, industry partners, and more, we can create a future where everyone with TSC can realize their full potential—no matter how complex their journeys are to get there. Join us at tscalliance.org or contact us at info@tscalliance.org.

TSC Alliance Leadership

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Paul Waxlax Denver, CO

Corporate Advisory Board

Wilfred Cooper, Sr., Honorary Chair Chairman

WNC & Associates

Julianne Moore, Honorary Chair

E. Martina Bebin, MD, MPA Co-Director

University of Alabama TSC Clinic

Rita DiDomenico Vice President MUFG Bank

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Director

NYU Epilepsy Center

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Laura S. Lubbers, PhD Chief Scientific Officer

CURE Epilepsy

Alan Marshall Senior Director, Customer

Engagement

Jazz Pharmaceuticals

Ted Mastroianni

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Vice President, International

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Grant Writer Consultant

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Vice President, Community

Programs

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Development Coordinator

Zoë Fuchs

Science Project Manager

Emebet G/Micheal Senior Accountant Jaye Isham

Senior Vice President, Strategic

Communications

Carly Kaye

Community Programs Manager

Dan Klein

Director, Digital Platforms

Justin Martucci

Development Systems Manager

Shelly Meitzler

Director, Community Education &

Resources

Lisa Moss

Vice President, Donor Relations

Jo Anne Nakagawa Director, Clinical Projects

Ashley Pounders, MSN, FNP-C Director, Medical Affairs

Gabrielle Rushing, PhD Director, Research

Gail Saunders Senior Associate Director, Community Programs

Katie Smith

Director, Government & Global

Affairs

Rachel Wojnilower Executive Assistant

Anne Wolfe

Senior Community Programs

Manager

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TSC Alliance Volunteer Leaders

2022 Community Alliances

The TSC Alliance's Community Alliance program fosters stronger links between the TSC Alliance and TSC communities and individuals across the United States. A Community Alliance is an informal, yet structured, group of empowered and caring volunteers who work closely with the national TSC Alliance to facilitate local connections for individuals and families affected by tuberous sclerosis complex (TSC), raise revenue and increase awareness while supporting the mission of the organization.

TSC Alliance of Alabama Margaret Cox, Chair margaretgcox@gmail.com

TSC Alliance of Alaska Rebecca Racenet, Chair rebecca.racenet@gmail.com Erica Chilla, Vice Chair ericachilla@gmail.com

TSC Alliance of Connecticut Jennifer Waldron, Chair jaw81873@gmail.com

TSC Alliance of Dallas/Fort Worth Leslie Russell, Chair leslie.russell2@verizon.net Lauren Sims, Vice Chair lsweldon@yahoo.com

TSC Alliance of Delaware/Lehigh Valley Liz Saadoun, Co-Chair fablizzy16@verizon.net Devon McCollum, Co-Chair devonmccollum@yahoo.com

TSC Alliance of Florida Amy Hightower, Co-Chair amynicolehightower@gmail.com

Vanessa Vazquez, Co-Chair vvazq004@fiu.edu

TSC Alliance of Georgia Wendi Scheck, Chair 5schecks@gmail.com

TSC Alliance of Greater Chicago Geri Greenberg, Chair tscgreaterchicago@icloud.com

TSC Alliance of Greater Kansas City Amy Dublinske, Chair dublinske@sbcglobal.net

TSC Alliance of Hawaii Trisha Kam, Chair trish.kam457@gmail.com TSC Alliance of Indiana Cassie Souder, Chair cassiesouder@gmail.com

TSC Alliance of the Intermountain West Kelli Sutherland, Co-Chair kell_corb@yahoo.com Brittani Rich, Co-Chair Brittanif81@gmail.com

TSC Alliance of Kentucky/West Virginia Melissa Bartley, Chair melissabartley1@gmail.com

TSC Alliance of Maryland Lauren Shillinger, Co-Chair lauren.shores@gmail.com Kristy Nunley, Co-Chair kjnunley@gmail.com

TSC Alliance of Michigan Mary Jo Cardona, Chair Micardona11@gmail.com

TSC Alliance of Nevada Kathleen Kingston, Co-Chair Ivkathyk@gmail.com Tara Zimmerman, Co-Chair taraczimmerman@gmail.com

TSC Alliance of New York/New Jersey Laura Marks, Co-Chair Ijmarks@gmail.com Rhiannon Schade, Co-Chair rhiannon.schade@gmail.com

TSC Alliance of North Carolina Karen Johnston, Chair karen.m.whittington@gmail.com Virginia Musselman, Vice Chair vmccray90@gmail.com

TSC Alliance of Northern California Lilian Ansari, Co-Chair Lilian@ansari.us.com Jennifer Yeamans, Co-Chair jennifer.yeamans@gmail.com

2022 World TSC Conference • July 28-31, 2022 • Dallas, Texas

TSC Alliance of Ohio Tammy Thorne, Chair tammyskouns@gmail.com

TSC Alliance of Oklahoma Heather Lens, Chair heather.lens@gmail.com

TSC Alliance of the Pacific Northwest Gloria Triebenbach, Chair batnblueyes2@aol.com Debbie Rothweiler, Vice Chair c6vett15@gmail.com

TSC Alliance of the Rocky Mountain Region Dana Holinka, Chair emme01@msn.com

TSC Alliance of South Carolina Amy Bredeson, Chair acbredeson@gmail.com

TSC Alliance of Southern California Tara Palage, Chair tara.palage@yahoo.com Lesley Holmes, Vice Chair lesleyholmes2015@gmail.com TSC Alliance of St. Louis/Southern Illinois Jackie Grenia, Chair jackiegrenia@yahoo.com

TS Alliance of the Upper Midwest Alanna Spotts, Chair alannaspotts@gmail.com

TSC Alliance of Washington, DC/Virginia Adrianne Cohen, Vice Chair adriannecohen@gmail.com

TSC Alliance of Western Pennsylvania Kelsey Hudson, Chair jjsmom0905@gmail.com

TSC Alliance of Wisconsin Kristin DeGueme, Chair kristin.degueme@gmail.com Nicole Galarmo, Vice Chair nicolegalarno@gmail.com

2022 Adult Regional Coordinators

Each Adult Regional Coordinator acts as a resource for adults to connect and support other adults with TSC. They are knowledgeable about resources, events, support meetings, conferences, and research that pertains to adults with TSC. The Adult Regional Coordinators also provide support to the Community Alliances to promote adult involvement in local communities, and they are involved with government relations, encouraging adult involvement and recruitment in the TSC Alliance's Government Action Team.

Region 1: Washington, Oregon, Idaho, Montana, Wyoming, Alaska

Rob and Shannon Grandia, supported by Daniel Price grandia@verizon.net danielnprice@gmail.com

Region 2: California, Arizona, Nevada, New Mexico, Utah Rob and Shannon Grandia, supported by Daniel Price grandia@verizon.net danieInprice@gmail.com

Region 3: Colorado, Nebraska, South Dakota, North Dakota, Iowa, Minnesota, Wisconsin Danielle Clark supported by Melyssa McDonough danielle.clark@hotmail.com melyssamcdonough@gmail.com

Region 4: Texas, Oklahoma, Kansas, Missouri, Arkansas, Louisiana Jake Irby supported by Emilie Peters and Foster Hyde Irby.iake@amail.com

emilie.peters97@gmail.com Fosterhyde07@gmail.com Region 5: Illinois, Michigan, Indiana, Ohio, Pennsylvania Phillip Tallman

Philip.tallman@gmail.com

Region 6: Tennessee, Kentucky, Georgia, Alabama, Mississippi, Florida Kate Crossett Kate.barbato@gmail.com

Region 7: Connecticut, Vermont, Maine, Rhode Island, New Hampshire, New York, New Jersey, Massachusetts Iris Mustich Irismustich@gmail.com

Region 8: Delaware, Virginia, West Virginia, Maryland, North Carolina, South Carolina, Washington DC Kathy Henkel uneakmom@gmail.com

Social Media: Danielle Clark danielle.clark@hotmail.com

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2022 Dependent Adult Transition Resource Coordinators

Dependent Adult Transition Resource Coordinators are knowledgeable in state resources, events, support meetings, conferences, and any new research on TSC that pertains to dependent adults and transitioning young adults with TSC and their caregivers. They work collaboratively with Community Alliances to support and connect dependent adults, transitioning young adults and their caregivers to state resources.

Alaska

Rebecca Racenet, rebecca.racenet@gmail.com

Alabama

Jessica White, jessiwhite.al.tsc@gmail.com

Arizona

Debora Moritz, deboramoritz@hotmail.com

California

Lilian Ansari, Lilian@ansari.us.com Faith Golden, Specialkids330@gmail.com Shannon Grandia, Grandia@verizon.net

Colorado

Dana Holinka, emme01@msn.com

Georgia

Reiko Donato, donator@bellsouth.net Guillermo Henrici, mhenrici@gmail.com

Hawaii

Cindy Fowler, chcinrox@aol.com

Illinois

Paula Krischel, krishchelp5@gmail.com

Illinois/Missouri

Rene Friedel, rfriedel@chartner.net

Kansas

Lisa Hart, Lisa.hart@usd253.net

Louisiana

Julie Comeaux, julie.tscawareness@gmail.com

Michigan

Treasa Bolger-Dunlap, mtbolger@comcast.net

Minnesota

Alice Hulbert, alihul@aol.com John Hulbert, jhulb33170@aol.com

Nevada

Kathy Kingston, lvkathyk@gmail.com

New York

Laura Marks, TSCMamaNYC@gmail.com

North Dakota

Kim Melhoff, kamelhoff@gmail.com

Ohio

Tammy Kouns, tammyskouns@gmail.com Nancy Weir, weirnancy@gmail.com

Oregon

Susan McBrine, susanmcbrine@gmail.com

Pennsylvania

Teresa O'Brien, tstopp1970@gmail.com

Texas

Laura Buckner, Laura.buckner@utexas.edu Kathy Groves, kathryn.a.groves@icloud.com Leslie Russell, leslie.russell2@verizon.net

Lucy Martinez, Lucy.martinez@encompasshealth.com

Virginia

Kathi Dills, kteach599@msn.com

Washington

Leslie Fox, leslie.a.fox@comcast.net

Wisconsin

Lori Denzine, Sportie883@hotmail.com

2022 Education Parent Mentors

The TSC Alliance developed the Educator Mentor Program (EMP) to support school systems in understanding the learning needs of children with TSC. The EMP is made up of administrators, therapists, psychologists and teachers from pre-kindergarten to college level having experience with TSC. They provide one-on-one support to any professional in the school system needing education and support in understanding the educational complexities of TSC.

Alaska

Rebecca Racenet, rebecca.racenet@gmail.com

Alabama

Jessica White, jessiwhite.al.tsc@gmail.com

California (Central)

Kaycee Kincade, Kaycee716@yahoo.com

California (Southern)

Faith Golden, Specialkids330@gmail.com

California (Northern)

Lilian Ansari, Lilian@ansari.us.com

Hawaii

Cindy Fowler, chcinrox@aol.com

Illinois

Theresa Conyers, Theresa.conyers@yahoo.com

Illinois/Missouri

Rene Friedel, rfriedel@charter.net

Iowa/Nebraska

Wendy Andersen, wendy@wendyandersencoaching.com

Kansas

Brooke Carpenter, brooke@becarpenter.com

Lisa Hart, Lisa.hart@usd253.net

Kentucky

Brianah Doeden, Brianah.doeden@protonmail.com

Louisiana

Julie Comeaux, julie.tscawareness@gmail.com

Michigan

Treasa Bolger-Dunlap, mtbolger@comcast.net

North Carolina

Amy Quintana, Amy.Quintana@me.com

Nancy Blankenship, nancyblankenship@icloud.com

North Dakota

Kim Melhoff, kamelhoff@gmail.com

New Jersey

Marilyn Pruce, Marilyn.pruce@gmail.com

New York

Blair Caddy, Blairc99@yahoo.com Naomi Haiztat, nhaiztat@gmail.com Laura Marks, TSCMamaNYC@gmail.com Daniela Fleisman, Daniela.fleisman@gmail.com

Ohio

Tammy Kouns, tammyskouns@gmail.com

Pennsylvania (East)

Kate Crossett, Kate.barbato@gmail.com

Pennsylvania (West)

Teresa O'Brien, tstopp1970@gmail.com

Texas

Leslie Russell, leslie.russell2@verizon.net

Virginia

Jill Fischer, jillwfischer@aol.com

National for Spanish Speakers

Vanessa Vazquez, vvazq004@fiu.edu

If you are interested in becoming a peer-to-peer support volunteer, please contact Shelly Meitzler, Director, Community Education & Resources, at smeitzler@tscalliance. org or (240) 638-4650.

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Special Events

Thank you to all who served as chairs and organizers for our recent special events—your dedication has helped to raise funds, increase awareness and provide community-building opportunities. If you are interested in serving on the National Walk Committee for Step Forward to Cure® TSC or as a special events leader in your community, please contact April Cooper, Vice President, Community Programs, at acooper@tscalliance.org or 301-562-9890.

Step Forward to Cure® TSC Walk Chairs, **National Planning Committee and Community Events**

Shalonda Alexander (TX), Lilian Ansari (CA), Laurisa Ballew (KS), Andrea Beebe (DE), Ryan Beebe (DE), Emily Berryman (KY), Treasa Bolger Dunlap (MI), Bridget Britt (CA), Donna Bullard (MO), Loren and Christy Buntrock (WI), Terri Butler (AZ), Mary Jo Cardona (MI), Michal Ceplar (DC), Joseph Cervantez (TX), Danielle Clark (TX), Bridgett Claxton Langstaff (NY), David Cowlin (IN), Margaret Cox (AL), Lori Distefano (NY), Jaime Edwards (MD), Alexandria Ewing (TN), Preston Fitzgerald (TX), Cindy Fowler (HI), Ashley Gabbard (OH), Jonathan Goldstein (NY), Shannon Grandia (CA), Heather Harden (MS), Jonathan Harden (MS), Frances Harper (CA), Amy Hightower (FL), Chelsea Holman (NV), Lesley Holmes (CA), Jill Hyman (CA), Kari Ihle (MN), Rachael Jackson (TX), Cassie Jimenez (TX), Jaime Jodoin (FL), Karen Johnson-Wenger (IL), Karen Johnston (NC), Amanda Keller (WI), Kathleen Kingston (NV), Jessica Krefting (AL), Jennifer Kuehn, Mary Ann Lamb (NC), Heather Lens (OK), Aida Lucero (CO), Fran Lyons (PA), Jack Lyons (PA), Katie Martin (CA), Diane McSwain (IL), Jessie Meyers, Keri Moore (MI), Debora Moritz (AZ), Heather Morris (NC), Nathalie Mortine (OH), Noor Panjwani (TX), Lauren Perry (TN), Lori Quirino (OH), Brittani Rich (UT), Mary Roberds (MD), Molly Russell (MI), Pat Schmutte (IN), Lauren Shillinger (DC), Sylvia Sparby (TX), Kirsten Swain (OK), Pam Sztukowski (WI), Jackie Tallman (IL), Phil Tallman (IL), Rob Thurston (UT), Gloria Triebenbach (WA), Jeff Triemer (GA), Vanessa Vazquez (FL), Stacie Verrill (NH), Jennifer Waldron (CT), Adrienne Wasserman (WA), Carol Ann White (NY), Jessica White (AL), Jacqueline Wolak (IL), Bill and Nancy Youmans (CO), Tara Zimmerman (NV), Keith Hall Golf Tournament - Family & Friends of Keith A. Hall (CT) and Carly Kaye Design Studio for Step Forward to Cure TSC shirts.

Comedy for a Cure® Planning Committee

Katrina Caban, Tiffani Goff (Auction Chair), Rob Grandia, Shannon Grandia, Christy Hobart, Lesley Holmes, Jill Hyman (Chair), Michelle Ingram-DeLong, Kent Kahlen, Cassie Kelle, Cathy Krinsky, Jeff Krol, Rose Krol, Judy LaGraff, Tara Palage, Mary Roberds, Henry Shapiro, Chris Sheffield, Patrick Sheffield, Annie Szilagyi, Lisa Szilagyi.

Minnesota Sound Bites Planning Committee

Carolyn Gilde, Janie Frost, Michael Giacomini, Chris Hawkey, Sarah Jane Hoey, Justin and Kari Ihle, Jessica Iserman, Catherine Jensen, Amber McClure, Darren Miles, Kat Perkins, Mary Roberds, Judy Shoulak, Alanna Spotts, Jeanne Theis, Angela Wiltscheck.



Global Alliances

Officially launched in 2014 with the TSC Alliance of Israel, the TSC Alliance Global Alliance Program was created as a way to address unmet needs within the global TSC community at the country level. This program provides the opportunity for the TSC Alliance to share experiences and assist in the start-up or support of TSC-related organizations in other countries.

A Global Alliance is a structured group of empowered and caring volunteers who work closely with the TSC Alliance to accomplish the following: facilitate local connections for individuals and families affected by TSC, build relationships with healthcare providers, raise revenue, and increase awareness while supporting the mission of the organization. The Global Alliance program forges stronger links between the TSC Alliance, communities, and individuals around the world.

Hungarian Foundation for Tuberous Sclerosis Magyar Sclerosis Tuberosa Alapítvány President: Imola Dankahàzi www.tsc.hu

Tuberous Sclerosis Alliance Foundation (Thailand)
Co-Founders: Carl and Nok Brenden

TSC Alliance of India President: Rahul Vipparthi www.tsa-india.org

TSC Alliance of Israel President: Viktor Perets www.tsc.org.il

TSC Alliance of Mexico President: Ary Agami www.facebook.com/tsallianceofmexico

Tuberous Sclerosis Canada Sclérose Tubéreuse (TSCST) Chair: Cathy Evanochko www.tscanada.ca

For more information on the Global Alliance Program and Global Partnerships, please contact Katie Smith, Director, Government and Global Affairs, at ksmith@tscalliance.org or (301) 562-9890.

About TSC International (TSCi)



Tuberous Sclerosis Complex International (TSCi) is a world-wide consortium of existing tuberous sclerosis complex associations and organizations, serving as an avenue to empower those affected by tuberous sclerosis complex (TSC), including individuals, families, caregivers, educators, and health care providers. In addition, TSCi provides a forum to share information, exchange ideas and methods, co-fund research projects and promote increased international awareness of TSC. Additional information can be found at www.tscinternational.org.

Europe

European Tuberous Sclerosis Complex Association (ETSC) Established in 2012, ETSC is a federation of Tuberous Sclerosis Associations.

ETSC believes in total "patient experience" and that interaction with all healthcare providers and relative agencies is paramount.

Website: www.e-tsc.eu Email: info@e-tsc.eu

Argentina

Asociación Argentina de Esclerosis Tuberosa (ARGET) Website: www.asociacionarget.blogspot.com.ar

Email: asociacionarget@gmail.com

Australia

Tuberous Sclerosis Australia (TSA) Website: www.tsa.org.au Email: info@tsa.org.au

Austria

Tuberöse Sklerose Complex Mitanand Website: www.tuberoesesklerose.at Email: info@tuberoesesklerose.at

Belgium

be-TSC

Website: www.betsc.be Email: info@betsc.be

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Brazil

Brazilian Association of Tuberous Sclerosis - Associação

Brasileira de Esclerose Tuberosa (ABET)

Website: www.abet.org.br Email: abetbh@gmail.com

Canada

Tuberous Sclerosis Canada Sclerose Tuberuse (TS Canada

ST)

Website: www.tscanada.ca Email: tscanadast@gmail.com

China

TSC China - 北京蝴蝶结结节性硬化症罕见病关爱中心

Email: info@tscchina.org

Denmark

Dansk Forening for Tuberøs Sclerose Heibergsvænge

Website: http://tsdanmark.dk Email: formand@tsdanmark.dk

Finland

TS-YHDISTYS

Website: www.tuberoosiskleroosi.fi

France

French TSC association - Association française Sclérose

tubéreuse de Bourneville (ASTB) Website: www.astb.asso.fr Email: contact@astb.asso.fr

Germany

German Tuberous Sclerosis Association (TSDe.V) -

Tuberöse Sklerose Deutschland e.V.

Website: www.tsdev.org Email: info@tsdev.org

Greece

Tuberous Sclerosis Association of Greece – Ελληνική

Εταιρεία Οζώδους Σκληρύνσεως (Ε.Ε.Ο.Σ.)

Website: www.tsahellas.gr Email: tsahellas@ath.forthnet.gr

Hungary

Hungarian Foundation for Tuberous Sclerosis

Website: www.tsc.hu Email: info@tsc.hu

Hong Kong

Tuberous Sclerosis Complex Association of Hong Kong -

香港結節性硬化症協會

Email: tscahk0515@gmail.com

India

TSC Alliance of India Website: www.tsa-india.org

Ireland

TSC Ireland

Website: www.tscireland.org Email: tscireland@outlook.com

Israel

TSC Alliance of Israel – סיזורלקס סורבוטל תילארשיה התומעה

Website: www.tsc.org.il Email: mail@tsc.org.il

Italy

Tuberous Sclerosis Association, Italy – Associazione

Sclerosi Tuberosa (AST, AST-ONLUS) Website: www.sclerosituberosa.org Email: info@sclerosituberosa.org

Japan

The Japanese Society of Tuberous Sclerosis Complex

Website: www.tsceres.org Email: tscres@juntendo.ac.jp

Macedonia

Daite ni krilia

Institute of Radiotherapy and Oncology Skopje

Website: www.dajtenikrilja.mk

Mexico

TSC Alliance of Mexico

Email: tsalliancemexico@gmail.com

Netherlands

Tuberous Sclerosis Netherlands Foundation – Stichting

Tubereuze Sclerosis Nederland (STSN)

Website: www.stsn.nl Email: info@stsn.nl

New Zealand

Tuberous Sclerosis Complex New Zealand (TSCNZ) Website: www.tsc.org.nz Email: info@tsc.org.nz

Norway

Norsk forening for Tuberøs Sklerose Website: www.nfts.no

Email: post@nfts.no

Poland

Stowarzyszenie Chorych na Stwardnienie Guzowate

Website: www.stwardnienie-guzowate.eu Email: stowarzyszenie.stw.guz@wp.pl

Portugal

The Portuguese Tuberous Sclerosis Association – Associação de Esclerose Tuberosa em Portugal (AETN)

Website: www.esclerosetuberosa.org.pt Email: info@esclerosetuberosa.org.pt

Russia

Association of patients with tuberous sclerosis – Ассоциация больных туберозным склерозом Website: epileptologhelp.ru/about/association-of-pa-

tients-with-tuberous-sclerosis

Email: tsc.rus@mail.ru

Serbia

Tuberous Sclerosis Association of Serbia – Udruženje za tuberoznu sklerozu Srbije

Email: 2013utss@gmail.com

Slovakia

ASTUS - Asociácia tuberóznej sklerózy (ASTUS, n.o.)

Website: www.diagnozatsc.sk

Email: astus@astus.sk or info@diagnozatsc.sk

Spain

Spanish Tuberous Sclerosis Association - Asociación

Nacional de Esclerosis Tuberosa Website: www.esclerosistuberosa.org Email: esclletuber10@gmail.com

South Africa

Tuberous Sclerosis South Africa (TSSA) Email: rehana.effendi@uct.ac.za

Sweden

TSC Sweden – TSC Sverige Website: http://tsc-sverige.se Email: annika@tsc-sverige.se

Switzerland

STB Suisse

Website: www.stbsuisse.ch Email: stbsuisse@postmail.ch

Taiwan

Taiwan Tuberous Sclerosis Complex Association (TTSC) – 台灣結節硬化症協會

Website: www.ttsc.org.tw Email: tscare@gmail.com

Thailand

Tuberous Sclerosis Alliance Foundation Email: tsalliancethai@gmail.com

Ukraine

Non-governmental Organization All-Ukrainian Aid Association for People with Tuberous Sclerosis website: www.tuberoussclerosis.com.ua

email: tscukraine@gmail.com

United Kingdom

Tuberous Sclerosis Association (TSA) Website: www.tuberous-sclerosis.org Email: social@tuberous-sclerosis.org

United States

TSC Alliance

Website: www.tscalliance.org Email: info@tscalliance.org

TSC

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TSC Clinics

The TSC Alliance supports the vital role a tuberous sclerosis complex (TSC) clinic can play in improving the lives of those affected. The TSC Alliance provides a link to a TSC Clinic's website, if available. Please contact the TSC Clinic or go to its website for details about their specialists, clinic services, billing and insurance.

The following information is current as of March 2021 and subject to change. For complete information on each clinic, visit www.tscalliance.org/tsc-clinics.

*Denotes a TSC Center of Excellence

ALABAMA

Birmingham

University of Alabama Birmingham*

ARIZONA

Phoenix

APNNA – Arizona Pediatric Neurology & Neurogenetics Associates

ARKANSAS

Little Rock

Arkansas Children's Hospital

CALIFORNIA

Loma Linda

Loma Linda University Health Care

Los Angeles

- Children's Hospital Los Angeles
- University of California Los Angeles*

Oakland

UCSF Benioff Children's Hospital Oakland

Orange

Children's Hospital of Orange County

San Diego

Rady Children's Hospital San Diego

Stanford

Lucile Packard Children's Hospital and Stanford Hospital and Clinics*

COLORADO

Aurora (Denver)

Children's Hospital Colorado and University Hospital

CONNECTICUT

Farmington

Connecticut Children's Medical Center

DELAWARE

Wilmington

Nemours/duPont Hospital for Children

DISTRICT OF COLUMBIA AREA

Washington, DC

Children's National Medical Center

FLORIDA

Gainesville

The University of Florida Health

Miam

Nicklaus Children's Hospital Miami

Orlando

- · Advent Health Medical Group Epilepsy at Orlando
- Orlando Health Arnold Palmer Children's Hospital

St. Petersburg

Johns Hopkins All Children's Hospital

Tampa

H. Lee Moffitt Cancer Center

GEORGIA

Atlanta

Children's Healthcare of Atlanta

ILLINOIS

Chicago

- University of Chicago Medical Center
- Ann & Robert H. Lurie Children's Hospital of Chicago
- · University of Illinois at Chicago

IOWA

Iowa City

University of Iowa Hospital & Clinics

KENTUCKY

Louisville

Novak Center/University of Louisville

MARYLAND

Baltimore

- Kennedy Krieger Institute
- University of Maryland Medical Center*

Rethesda

Mid-Atlantic Epilepsy and Sleep Center

MASSACHUSETTS

Boston

- Massachusetts General Hospital*
- Boston Children's Hospital*

MICHIGAN

Detroit

Children's Hospital of Michigan

Grand Rapids

Helen DeVos Children's Hospital

Royal Oak

Beaumont Children's Hospital

MINNESOTA

Roseville

Upper Midwest TSC Clinic Without Walls*

MISSOURI

Kansas City

Children's Mercy Hospital

St. Louis

- Cardinal Glennon Children's Hospital, Saint Louis University School of Medicine
- Washington University*

NEW JERSEY

Hackensack

Northeast Regional Epilepsy Group

Livingston

Institute of Neurology at St. Barnabas

NEW YORK

Bronx

Montefiore Medical Center

Hawthorne, Westchester, and Hudson Valley Clinic Without Walls serving NYC, Westchester, Hudson Valley & Connecticut

New York City

- Columbia University Irving Medical Center/New York Presbyterian
- New York University Langone Medical Center

NORTH CAROLINA

Charlotte

Atrium Healthcare at Levine Children's Hospital

Durham

Duke Medical Center*

Winston-Salem

Wake Forest Baptist Medical Center

OHIO

Akron

Akron Children's Hospital

Cincinnati

Cincinnati Children's Hospital*

Cleveland

- The Cleveland Clinic
- Rainbow Babies and Children's Hospital, University Hospitals of Cleveland Medical Center

Columbus

Nationwide Children's Hospital

Dayton

Dayton Children's Hospital

PENNSYLVANIA

Hershey

Penn State Health/Hershey Medical Center

Philadelphia

The Hospital of the University of Pennsylvania and Children's Hospital of Philadelphia

Pittsburgh

Children's Hospital of Pittsburgh

SOUTH CAROLINA

Columbia

Prisma Health

TENNESSEE

Memphis

Le Bonheur Children's Hospital*

Nashville

Monroe Carell, Jr. Children's Hospital at Vanderbilt

TEXAS

Austin

Dell Children's Hospital

Dallas

Scottish Rite for Children

Fort Worth

Cook Children's Hospital

Houston

- McGovern Medical School, University of Texas Health Science Center at Houston*
- Texas Children's Hospital affiliated with Baylor College of Medicine*

UTAH

Salt Lake City

University of Utah Primary Children's Hospital

VIRGINIA

Charlottesville

The University of Virginia

Richmond

Virginia Commonwealth University Health System

WASHINGTON

Seattle

- · Seattle Children's Hospital
- Swedish Neuroscience Institute

WISCONSIN

Milwaukee

Children's Hospital of Wisconsin

CANADA

Montreal, Quebec

- CHU Sainte-Justine, University of Montreal
- Centre Hospitalier de l'Université de Montréal

Toronto, Ontario

- The Hospital for Sick Children
- University Health Network

INDIA

Mumbai, Maharashtra

SRCC Children's Hospital by NH

ISRAEL

Be'er Shiva

Soroka University Medical Center

Jerusalem

Hadassah Medical Center

Tel Hashomer

Sheba Medical Center

MEXICO

Merida, Yucatan

Neural Clinic Mexico

Monterrey, Nuevo León

Hospital Universitario "José Eleuterio González"

Universidad Autónoma de Nuevo León

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Session Descriptions

TSC 101 Part I - For Newly Diagnosed Individuals or Families

Discussion - Edelweiss, Thursday, 4:00-5:00 p.m.

Opening Dinner

Dinner - Imperial Ballroom, Thursday, 6:30–8:30 p.m.

A welcome to the 2022 World TSC Conference and Dallas during the first gathering of all attendees. The dinner presentaion will include what to expect of the conference and highlights of local Dallas "must see" attractions.

Opening General Session: Global Collaborations (TANDem) and Consensus Guidelines Updated

Lecture - Ballroom DE, Friday, 8:30-10:00 a.m.

The conference's opening session will showcase the powerful impact achieved through international collaborations: (1) recently published updated global consensus guidelines for diagnosis, surveillance, and treatment of TSC, and (2) the TAN-Dem project, empowering people living with TSC to learn to identify TSC-associated neuropsychiatric disorders (TAND) and thereby benefit from timely support.

* Navigating the Complexities for Special Needs Care in TSC (SSI, Waivers, Medicaid, Access, Financial Planning and Guardianship)

Panel - Ballroom A, Friday, 10:30 a.m.-12:00 p.m.

This panel session will discuss the transition from childhood to adulthood and the services and support needed for dependent adults. The parents or guardians of dependent adults or future dependent adults need to address the future financial and daily living needs of the dependent adult. This session will provide information and resources to help prepare for these future needs and provide guidance on timelines and common issues around these processes.

Using Technology to Your Advantage: Seizure Monitoring, Empowering Communication for the Nonverbal, and Futuristic Medical Care

Specialty - Ballroom B, Friday, 10:30 a.m.-12:00 p.m.

This session will review how technology can enhance communication and empower individuals to manage their health, now and in the future.

Seizure Types and Treatments

Lecture - Ballroom DE, Friday, 10:30 a.m.-12:00 p.m.

This session will address seizure types across all ages and will include a discussion of treatments available.

Organizing Your Thoughts and Emotions and Managing Relationships (TSC individual)

Workshop - Plum Blossom, Friday, 10:30 a.m.-12:00 p.m.

This session features TSC Alliance Adult Regional Coordinators and clinical experts discussing personal and professional experiences related to managing thoughts, emotions and relationships for adults impacted by TSC. The panel will also provide insights and resources on executive functioning and tools to help navigate conversations and relationships.

Preparing Your Family for Epilepsy Surgical Evaluation and Surgery

Discussion – Edelweiss, Friday, 10:30 a.m.–12:00 p.m.

TSC 101 Part II - Caregiver Guide to Care for your Loved One for Newly Diagnosed

Lunch & Learn - Plum Blossom, Friday, 12:00-1:00 p.m.

^{*}All are welcome; however, these sessions may be most relevant to individuals living in the United States.

Caregiver Mental Health and Wellbeing

Lunch & Learn - Ballroom A, Friday, 12:00-1:00 p.m.

Surgical Approaches and Assessments for Epilepsy and SEGA

Panel – Ballroom A, Friday, 1:00–2:30 p.m.

This panel will discuss different surgical interventions for epilepsy and SEGAs in children.

Accessing Managed and Palliative Care Throughout Your TSC Journey

Specialty - Ballroom B, Friday, 1:00-2:30 p.m.

This session will provide an overview of different supportive care across all ages and manifestations of TSC from experts in TSC, LAM, and palliative care.

Skin and Dental Manifestations and Treatment Options

Lecture – Ballroom DE, Friday, 1:00–2:30 p.m.

This lecture will address skin and dental manifestations of TSC, and how to treat them.

Preparing for Transitions - Childhood to Adulthood (Medical Care)

Workshop - Plum Blossom, Friday, 1:00-2:30 p.m.

This workshop will explain the difference between pediatric and adult medical care and provide strategies and tools to help prepare for your loved one's transition from a family–centric to a person–centric medical care model.

Women's Health Issues for TSC Adults

Discussion - Edelweiss, Friday, 1:00-2:30 p.m.

Accessing Early Childhood Services and Non-Medical Therapies

Panel - Ballroom A, Friday, 3:00-4:30 p.m.

This panel session will discuss the importance of accessing early intervention and preschool services, and the non–medical therapies including physical therapy, occupational therapy and speech and language therapy.

LAM, Treatment Options and Clinical Trials

Specialty - Ballroom B, Friday, 3:00-4:30p.m.

This session addresses the link between TSC and lymphangioleiomyomatosis (LAM). Learn about LAM treatment options, available clinical trials for individuals with LAM, and resources available for support through the LAM Foundation.

Genetics Made Simple

Lecture – Ballroom DE, Friday, 3:00–4:30 p.m.

The session will simplify the genetics of TSC into digestible, easy-to-understand information.

TAND - Aggressive Behavior Management

Workshop – Plum Blossom, Friday, 3:00–4:30 p.m.

This workshop will provide practical tools & tips to manage aggressive & harmful behavior. We recognize how difficult these behaviors are to deal with at any age and the impact behavior has on accessing long term supports.

Men's Health Issues for TSC Adults

Discussion - Edelweiss, Friday, 3:00-4:30 p.m.

General Q&A, Panel of Day 1 Presenters

Lecture - Ballroom DE, Friday, 4:45-5:30 p.m.

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Reunión Para Familias Hispanohablantes - Spanish Education Meeting and Gathering

Workshop – Plum Blossom, Friday, 5:30–7:00 p.m.

Esta sesión proporcionará recursos y actualizaciones a las personas hispanohablante de la comunidad de TSC sobre información nueva en las guías clínicas de consenso para el tratamiento y la atención de las personas afectadas por TSC. La sesión se llevará a cabo en español y los asistentes tendrán la oportunidad de hacer preguntas a los panelistas expertos.

(This session will provide resources and updates to the Spanish–speaking TSC community on the revised clinical consensus guidelines for the treatment and care of individuals impacted by TSC. The session will be conducted in Spanish and attendees will have opportunities for Q&A with the expert panelists.)

General Session: TSC Community Driven Research Network (TSC Alliance Research Model)

Lecture - Ballroom DE, Saturday, 8:30-10:00 a.m.

This session will showcase the impact and vision of the TSC Alliance's research and government advocacy programs, and how that vision is shaped and achieved in partnership with the TSC community.

Sibling Workshop

Group - Cooper, Friday 10:30 a.m. - 12:00 p.m.

This moderated session for siblings will be an open forum for participants to share with each other.

TAND - ADHD, Anxiety, Autism (Research)

Panel - Ballroom A, Saturday, 10:30 a.m.-12:00 p.m.

This panel will discuss attention deficit hyperactivity disorder (ADHD), anxiety, autism, and the breakthrough research being conducted to better understand how it specifically affects individuals with TSC.

Caregiver Resources - Self and Respite Care

Specialty - Ballroom B, Saturday, 10:30 a.m.-12:00 p.m.

This session will address resources and strategies for managing the stress TSC can bring to your life, including tactics for improving your physical and mental health, small steps of self–care for your daily life and information on accessing respite care.

Kidney Tumors, Cysts and Treatment Options

Lecture - Ballroom DE, Saturday, 10:30 a.m.-12:00 p.m.

This lecture will focus on kidney issues and treatment options across all ages.

Seizure Action Plans and Rescue Meds

Workshop – Plum Blossom, Saturday, 10:30 a.m.–12:00 p.m.

In this workshop, you will hear about the importance of creating a Seizure Action Plan for you and/or your family member, including the use of rescue medications and how to manage emergencies. Additionally, the session will cover how to communicate the content of a Seizure Action Plan with educational accommodation teams and caregivers, as well as in the workplace. You will leave with the start of an actionable plan.

* Navigating Education System for a High School Certificate

Discussion - Edelweiss, Saturday, 10:30 a.m.-12:00 p.m.

Government Advocacy

Lunch & Learn - Ballroom B, Saturday, 12:00-1:00 p.m.

Peer-to-Peer Support Programs

Lunch & Learn - Ballroom A, Saturday, 12:00-1:00 p.m.

*All are welcome; however, these sessions may be most relevant to individuals living in the United States.

Special Events and Fundraising

Lunch & Learn - Plum Blossom, Saturday, 12:00-1:00 p.m.

Genetics, Reproductive Issues and Impact on Relationships

Panel – Ballroom A, Saturday, 1:00-2:30 p.m.

This session will discuss TSC genetics as it relates to relationships, including potential reproductive issues or choices.

Refractory Epilepsy - Now What?

Specialty - Ballroom B, Saturday, 1:00-2:30 p.m.

Epilepsy in TSC is often refractory. Seizures may not be controlled with initial therapies, resulting in the need to be aware of all possible therapeutic options to treat seizures in a person with TSC. This session will explore other treatment options neurologists may discuss with families and individuals with TSC of any age to try to bring seizures under control.

Access to TSC Clinical Care (with international component)

Lecture - Ballroom DE, Saturday, 1:00-2:30 p.m.

This lecture will provide an overview of issues related to accessing medicines and clinical care worldwide. We will discuss major barriers to affordable access across different countries and regions, the progress that has been made by TSC organizations around the world, and opportunities for global collaboration to improve access to care.

Advocacy in the Education System at All Levels

Workshop - Plum Blossom, Saturday, 1:00-2:30 p.m.

This workshop session will provide an understanding of the IEP or 504 plans, Goals, Progress and Transitions. The session provides tools and insight into the importance of advocating for your child's education at any level.

TAND - Mental Health Issues in Living with TSC as an Independent and Semi-Independent Adult

Discussion – Edelweiss, Saturday, 1:00–2:30 p.m.

* Finding Your Way Home - Creative Housing Options / Microboards

Panel - Ballroom A, Saturday, 3:00-4:30 p.m.

This panel session will discuss housing options for Dependent and Semi-Independent TSC Adults that incorporate inclusion in the community based on the unique needs of the individual.

Managing Progressive Kidney Disease

Specialty - Ballroom B, Saturday, 3:00-4:30 p.m.

This session will focus on progressive kidney disease in individuals affected by TSC and the treatment options available to protect kidney function over time.

Gene Therapy - Hopes and Hurdles

Lecture - Ballroom DE, Saturday, 3:00-4:30 p.m.

This session will explore the possibilities for gene therapy in TSC and the challenges researchers face to make it a reality. Although gene therapy as a cure for TSC – or for most diseases – remains far away, scientists continue to find new ways to tackle the challenges and bring gene therapy closer to being a viable treatment.

TAND - Feeding, Elopement and Potty Training

Workshop – Plum Blossom, Saturday, 3:00–4:30 p.m.

This interactive workshop will provide examples of a behavior action plan to develop in the areas of feeding, bathroom, and issues around elopement (wandering off or escaping) by board certified behavior analysts.

*All are welcome; however, these sessions may be most relevant to individuals living in the United States.

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What's Next? Tools to Empower You on Your Medical Journey to an Adult Neurologist

Discussion – Edelweiss, Saturday, 3:00–4:30 p.m.

General Q&A, Panel of Day 2 Presenters

Lecture - Ballroom DE, Saturday, 4:45-5:30 p.m.

Other Organ Manifestations - Liver, Pancreas, Heart and Eyes

Panel - Ballroom A, Sunday, 8:30-10:00 a.m.

This session will discuss tumors associated with TSC including liver and retinal hamartomas, pancreatic neuroendocrine tumors, and non–malignant tumors in the heart. The panelists will also provide general recommendations on monitoring and treating these tumors.

Transition into Adulthood – Employment and Higher Education

Specialty - Ballroom B, Sunday, 8:30-10:00 a.m.

This session will provide insights and resources for young adults impacted by TSC and their families to approach the challenges of transitions to adulthood. The group will share their personal experiences and key considerations for semi–independent and independent individuals impacted by TSC who are transitioning out of secondary school and into higher education, vocational training, or employment.

TAND - Behavioral Interventions and Overview of Medications

Lecture - Ballroom DE, Sunday, 8:30-10:00 a.m.

This session will focus on behavioral issues that are commonly associated with TSC. Discussion will ensue on behavioral/psychiatric conditions, identifying provider(s) for treatment, medical management of behavioral issues, and prevention/intervention tips.

* Navigating Health Care System to Better Access Medication

Workshop - Plum Blossom, Sunday, 8:30-10:00 a.m.

This interactive workshop will provide guidance and strategies on accessing medication and clinical resources and services in the US.

Sexual Development for Dependent and Semi-Independent TSC Adults

Discussion - Edelweiss, Sunday, 8:30-10:00 a.m.

Closing Session: Driving the Future of TSC - Clinical Trials and Clinical Care

Lecture - Ballroom DE, Sunday, 10:30 a.m.-12:00 p.m.

The impact of TSC research is realized by improving clinical care and outcomes in people living with TSC. This session will conclude the 2022 World TSC Conference by examining how the TSC community is vital to designing and carrying out clinical trials to identify new treatments to improve the lives of those affected by TSC.

*All are welcome; however, these sessions may be most relevant to individuals living in the United States.

Full Conference Agenda

Please note: TAND stands for TSC-Associated Neuropsychiatric Disorders.

*All are welcome; however, these sessions may be most relevant to individuals living in the United States.

Wednesday, July 27, 2022

6:30 p.m.-9:00 p.m. TSCi Reception, by invitation only, Fleur de Lis A & B

Thursday, July 28, 2022

8:00 a.m.-2:00 p.m. Nursing Symposium, by invitation only, Plum Blossom

Sponsored by: Pediatric Epilepsy Research Foundation

9:00 a.m.-2:00 p.m. TSCi Workshop, by invitation only, Fleur de Lis A & B

12:00 p.m.-5:00 p.m. Biosample Repository Blood Draw, Inverness

12:00 p.m.-6:00 p.m. General Conference Registration and Day Camp Registration, Conference Registration Room

and Atrium II

12:00 p.m.-6:00 p.m. TSC Store Open, outside Grand Ballrooms

2:30 p.m.-4:00 p.m. TSC Clinic Directors Meeting, by invitation only, Plum Blossom

3:00 p.m.-6:00 p.m. Exhibit Hall Set Up, Grand Ballroom C

3:00 p.m.-5:00 p.m.

Meet-and-Greet Opportunities

Adults with TSC, Ballroom A

Hosts: Derek Bauer, MD; Rob and Shannon Grandia; Melyssa McDonough, MS, BCBA;

Mary Ann Scroggins Sponsored by: Ray Chan

· Adult Siblings, Gossip Bar

Hosts: Jocelyn Cenna, Carlie Russell

· Grandparents, CounterOffer

Host: Mary Ann Lamb

International Attendees, Fleur de Lis A & B

Hosts: Carla Fladrowski; Sergiusz Jóźwiak, MD, PhD; Chris Kingswood, MD;

Micaela Rozenberg

· Parents of Adults with TSC, CounterOffer

Hosts: April Cooper; John Hulbert, MD; Leslie Russell; Lisa Szilagyi

• Parents/Caregivers of Transitioning Young Adults, Jade Room

Hosts: Lisa Carlton, PhD; Steve Peters

Sponsored by: DiRx

• Parents/Caregivers of Young Children with TSC (ages 0-15), East Atrium

Hosts: Andrea Beebe, Lesley and Alex Holmes, Phil Tallman

Sponsored by: TotalCare RX and Perfecting Peds

• Social Networking Groups (Inspire, Instagram, Facebook), Jade Pavilion

Hosts: Danielle Clark, Carly Kaye, Sean Shillinger

Teens and Young Adults (ages 13-27), Obelisk A & B

Hosts: Preston Fitzgerald, Jonathan Goldstein, Rylee Grandia, Celie Johnson,

Kirk McCreary, Emily Racenet

Sponsored by: UCB, Inc.

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Thursday (cont.)

4:00 p.m.-5:00 p.m. TSC 101 for Newly Diagnosed Individuals or Families, Edelweiss

Moderator: Debora Moritz

Speakers: Angela Peron, MD; Rajsekar Rajaraman, MD

Sponsored by: Mallinckrodt

6:30 p.m.-8:30 p.m. Conference Welcome and Opening Dinner, Imperial Ballroom

TSC Alliance President and CEO Kari Luther Rosbeck; TSC Alliance Board Chair Peter Crino, MD, PhD; and Local TSC Clinic Directors Steven Sparagana, MD and

M. Scott Perry, MD

Sponsored by: Friends of Eric Parkes

8:30 p.m. Gathering for International Attendees, Media Grill + Bar 8:30 p.m. Adults with TSC Evening Activity: Bingo, Plum Blossom

Adults with TSC Evening Activity: Bingo, Plum Blossom
Event Leaders: Danielle Clark, Jonathan Goldstein, Shannon Grandia

8:30 p.m. TSC Dad's Outing, Gossip Bar

Event Leader: Sean Shillinger

8:30 p.m.-11:00 p.m. Parents and Dependent Adults Social Gathering, Stemmons

8:30 p.m.–11:00 p.m. **Teen Lounge Open**, *Obelisk A & B*

Friday, July 29, 2022

7:00 a.m.–8:30 a.m. Breakfast, Chantilly Ballroom

7:00 a.m.–8:30 a.m. Biosample Repository Blood Draw, Inverness

7:00 a.m.–8:30 a.m. Research Room Open, Fleur de Lis A & B

7:30 a.m.-5:30 p.m. Day Camp, Atrium I - Check-In: Stemmons Ballroom

8:00 a.m.–5:00 p.m. Exhibit Hall and TSC Resource Center, Grand Ballroom C

8:00 a.m.–5:00 p.m. TSC Store Open, outside Grand Ballrooms

8:00 a.m.-12:00 p.m. Conference Registration, Conference Registration Room and Atrium II

8:30 a.m.-10:00 a.m. OPENING GENERAL SESSION: Global Collaborations (TANDem) and Updated Consensus

Guidelines, Grand Ballroom D & E

Welcome: Peter Crino, MD, PhD, Chair, TSC Alliance Board of Directors

Moderator: Pradnya Gadgil, MBBS, MRCPCH

Family Speaker: Eva Schoeters

Speakers: Petrus de Vries, MBChB, FRCPsych, PhD; Hope Northrup, MD

Sponsored by: Upsher-Smith Laboratories, LLC

10:00 a.m.-10:30 a.m. **BREAK**

10:00 a.m.–3:00 p.m. Biosample Repository Blood Draw, Inverness

10:00 a.m.–3:00 p.m. Research Room Open, Fleur de Lis A & B

Friday (cont.)

10:30 a.m.-12:00 p.m. First Break Out Session

*PANEL: Navigating the Complexities for Special Needs Care in TSC (SSI, Waivers, Medicaid,

Access, Financial Planning and Guardianship), Grand Ballroom A

Moderator: Lisa Szilagyi

Panelists: John Hulbert, MD; Kelly Piacenti; Nadia Vann, JD

Sponsored by: MassMutual SpecialCare

SPECIALTY: Using Technology to Your Advantage: Seizure Monitoring, Empowering Communication for the Nonverbal, and Futuristic Medical Care, *Grand Ballroom B*

Moderator: M. Scott Perry, MD

Speakers: Debora Moritz, Rob Moss, Rahul Vipparthi

Sponsored by: Neurelis

LECTURE: Seizure Types and Treatments, Grand Ballroom D & E

Moderator: Rohini Coorg, MD

Speakers: Mark Keezer, MDCM, PhD; Elizabeth Thiele, MD, PhD; James Wheless, MD

Sponsored by: Mallinckrodt and Upsher-Smith Laboratories, LLC

WORKSHOP: Organizing Your Thoughts and Emotions and Managing Relationships

(TSC individual), Plum Blossom Moderator: Mary Kay Koenig, MD

Adult with TSC Panelists: Danielle Clark, Emilie Hyde, Foster Hyde, Phil Tallman

Speakers: Jamie Capal, MD; Petrus de Vries, MBChB, FRCPsych, PhD

DISCUSSION: Preparing Your Family for Epilepsy Surgical Evaluation and Surgery, Edelweiss

Moderator: Brenda Porter, MD, PhD

Speakers: Bridget Britt; Gerald Grant, MD; Dave and Rachel Johnson

12:00 p.m.–1:00 p.m. **Box Lunches**, *Ballroom Foyer*

LUNCH AND LEARN: TSC 101 Part II - Caregiver Guide to Care for Your Loved One for

Newly Diagnosed, Plum Blossom

Panelists: Debora Moritz, Kelly Piacenti, Leslie Russell

Sponsored by: Mallinckrodt

LUNCH AND LEARN: Caregiver Mental Health and Wellbeing, Ballroom A

Moderator: Tanjala Gipson, MD

Panelists: David Dunn, MD; Carla Fladrowski; Micaela Rozenberg

Sponsored by: Ovid Therapeutics

12:00 p.m.-1:00 p.m. Global Alliance Meeting, by invitation only, Lalique

12:00 p.m.–5:00 p.m. **Teen Lounge Open**, *Obelisk A & B*

1:00 p.m.-2:30 p.m. Second Break Out Session

^{*}All are welcome; however, these sessions may be most relevant to individuals living in the United States.

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Friday (cont.)

PANEL: Surgical Approaches and Assessments for Epilepsy and SEGA, Grand Ballroom A

Moderator: Dan Curry, MD

Panelists: Jeffrey Blount, MD; Asim Choudhri, MD; Gerald Grant, MD; Howard Weiner, MD

SPECIALTY: Accessing Managed and Palliative Care Throughout Your TSC Journey,

Grand Ballroom B

Moderator: Ashley Pounders, MSN, FNP-C

Speakers: Sarah Friebert, MD; Nishant Gupta, MD; Joshua Samuels, MD, MPH

LECTURE: Skin and Dental Manifestations and Treatment Options, Grand Ballroom D & E

Moderator: Klaus Werner, MD, PhD

Speakers: Elizabeth Gosnell, DMD, MS; Joyce Teng, MD, PhD

Sponsored by: Nobelpharma and Chemistry Rx

WORKSHOP: Preparing for Transitions - Childhood to Adulthood (Medical Care), Plum Blossom

Moderator: Michael Frost, MD

Speakers: Peter Crino, MD, PhD; Mustafa Sahin, MD, PhD; Rebecca Schultz, PhD, APRN, CPNP;

Lisa Szilagyi

Sponsored by: Horizon Therapeutics

DISCUSSION: Women's Health Issues for TSC Adults, Edelweiss

Moderator: Brenda Porter, MD, PhD

Speakers: Danielle Clark; Lara Friel, MD, PhD; Lisa Henske, MD; Hope Northrup, MD; Emily Phan

2:30 p.m.-3:00 p.m. BREAK

3:00 p.m.-4:30 p.m. Third Break Out Session

PANEL: Accessing Early Childhood Services and Non-Medical Therapies, Grand Ballroom A

Moderator: Rohini Coorg, MD

Panelists: Joanna Lomas-Mevers, PhD, BCBA; Shelly Meitzler, Ashley Pounders, MSN, FNP-C;

Kimberly Street, LMSW, LPN, CCM

SPECIALTY: LAM, Treatment Options and Clinical Trials, Grand Ballroom B

Moderator: Susan Sherman, MHA

Speakers: Nishant Gupta, MD; Lisa Henske, MD; Joel Moss, MD, PhD

LECTURE: Genetics Made Simple, Grand Ballroom D & E

Moderator: Steven Sparagana, MD

Speakers: Jennifer Glass, MS, CGC, LGC; Hope Northrup, MD; Zacil-Ha Vilchis Zapata, MD

WORKSHOP: TAND - Aggressive Behavior Management, Plum Blossom

Moderator: Peter Davis, MD

Speakers: Nathan Call, PhD, BCBA-D; David Dunn, MD; Tanjala Gipson, MD; Leslie Russell

DISCUSSION: Men's Health Issues for TSC Adults, Edelweiss

Moderator: Steve Roach, MD

Speakers: Derek Bauer, MD; Rob Grandia; Billy Holcombe, PhD

4:30 p.m.-4:45 p.m. **BREAK**

4:45 p.m.–5:30 p.m. GENERAL QUESTIONS AND ANSWERS (Panel of Presenters), Grand Ballroom D & E

Moderator: Kari Luther Rosbeck

Panelists: Nathan Call, PhD, BCBA-D; Peter Crino, MD, PhD; Dan Curry, MD; Tanjala Gipson, MD; Nishant Gupta, MD; Chris Kingswood, MD; Hope Northrup, MD; Joshua Samuels, MD, MPH;

Joyce Teng, MD, PhD; Howard Weiner, MD; James Wheless, MD

Friday (cont.)

5:30 p.m.-7:00 p.m. Reunión Para Familias Hispanohablantes - Spanish Education Meeting and Gathering,

Plum Blossom

Moderator: Darcy Krueger, MD, PhD

Speakers and Event Leaders: Ary Agami; Joao García Martínez, MD; Vanessa Vazquez;

Zacil-Ha Vilchis Zapata, MD Sponsored by: UCB, Inc.

6:30 p.m.–8:00 p.m. Dinner, Chantilly Ballroom

8:30 p.m. Adults with TSC Evening Activity: Karaoke, Plum Blossom

Event Leaders: Rob Grandia, Shannon Grandia, Daniel Price

8:30 p.m. Multicultural Social, for people interested in multi-cultural issues, Gossip Bar

Hosts: Lisa Carlton, PhD; Danielle Clark; Latrice Hamilton; Vanessa Vazquez

8:30 p.m.-11:00 p.m. Parents and Dependent Adults Social Gathering, Stemmons

8:30 p.m.–11:00 p.m. **Teen Lounge Open**, *Obelisk A & B*

8:45 p.m. Movie Night: Encanto, Jade Oval Lawn

Snacks available for purchase.

Saturday, July 30, 2022

7:00 a.m.-8:30 a.m. Breakfast, Chantilly Ballroom

7:00 a.m.–8:30 a.m. Biosample Repository Blood Draw, Inverness

7:00 a.m.–8:30 a.m. Research Room Open, Fleur de Lis A & B

7:30 a.m.–5:30 p.m. Day Camp, Atrium I – Check-In: Stemmons Ballroom

8:00 a.m.-10:00 a.m. Conference Registration, Conference Registration Room and Atrium II

8:00 a.m.-5:00 p.m. Exhibit Hall and TSC Resource Center, Grand Ballroom C

8:00 a.m.–5:00 p.m. TSC Store Open, outside Grand Ballrooms

8:30 a.m.-10:00 a.m. GENERAL SESSION: TSC Community Driven Research Network (TSC Alliance Research

Model), Grand Ballroom D & E Moderator: Martina Bebin, MD, MPA Family Speaker: Latrice Hamilton

Speakers: Dean Aguiar, PhD; Sara Chieffo; Zoë Fuchs; Steve Roberds, PhD; Gabrielle Rushing, PhD

Sponsored by: TSC Alliance Board of Directors

10:00 a.m.-10:30 a.m. BREAK

Sponsored by: Noble International

10:00 a.m.-3:00 p.m. Biosample Repository Blood Draw, Inverness
10:00 a.m.-3:00 p.m. Research Room Open, Fleur de Lis A & B

10:30 a.m.-12:00 p.m. Fourth Break Out Session

PANEL: TAND - ADHD, Anxiety, Autism (Research), Grand Ballroom A

Moderator: Mustafa Sahin, MD, PhD

Panelists: Nathan Call, PhD, BCBA-D; Jamie Capal, MD; Anna Jansen, MD, PhD;

Shafali Jeste, MD; Melyssa McDonough, MS, BCBA

Sponsored by: Psychogenics

^{*}All are welcome; however, these sessions may be most relevant to individuals living in the United States.

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Saturday (cont.)

SPECIALTY: Caregiver Resources - Self and Respite Care, Grand Ballroom B

Moderator: Debora Moritz

Speakers: Petrus de Vries, MBChB, FRCPsych, PhD; Kelly Piacenti; Karina Stormfeldt Helboe;

Kimberly Street, LMSW, LPN, CCM Sponsored by: MassMutual Special*Care*

LECTURE: Kidney Tumors, Cysts and Treatment Options, Grand Ballroom D & E

Moderator: Chris Kingswood, MD

Speakers: John Bissler, MD; Oded Volovelsky, MD, PhD

WORKSHOP: Seizure Action Plans and Rescue Meds, Plum Blossom

Moderator: Michael Frost, MD

Speakers: Peter Davis, MD; Pradnya Gadgil, MBBS, MRCPCH; Rob Moss; Rebecca Schultz,

PhD, APRN, CPNP

Sponsored by: Neurelis and Noema

*DISCUSSION: Navigating Education Systems for a High School Certificate, Edelweiss

Moderator: Shannon Grandia

Speakers: Kate Crossett, Shelly Meitzler

GROUP SESSION: Sibling Workshop, Cooper

12:00 p.m.–1:00 p.m. Box Lunches, Ballroom Foyer

LUNCH AND LEARN: Government Advocacy, *Ballroom B* Moderator: Sara Chieffo, Debora Moritz, Katie Smith

Sponsored by: BIO

LUNCH AND LEARN: Peer-to-Peer Support Programs, Ballroom A

Moderator: Shelly Meitzler

LUNCH AND LEARN: Special Events and Fundraising, Plum Blossom

Moderators: April Cooper, Gail Saunders

12:00 p.m.–5:00 p.m. Teen Lounge Open, Obelisk A & B

1:00 p.m.-2:30 p.m. Fifth Break Out Session

PANEL: Genetics, Reproductive Issues and Impact on Relationships, Grand Ballroom A

Moderator: Josh Samuels, MD, MPH

Panelists: Lara Friel, MD, PhD; Jennifer Glass, MS, CGC, LGC; Angela Peron, MD

SPECIALTY: Refractory Epilepsy - Now What?, Grand Ballroom B

Moderator: James Wheless, MD

Speakers: Peter Crino, MD, PhD; Sergiusz Jóźwiak, MD, PhD; Elizabeth Thiele, MD, PhD;

Michael Wong, MD, PhD

Sponsored by: Marinus Pharmaceuticals and Neurelis

LECTURE: Access to TSC Clinical Care, Grand Ballroom D & E

Moderator: Steve Roach, MD

Panelists: Carla Fladrowski; Pradnya Gadgil, MBBS, MRCPCH; Joao García Martínez, MD;

Darcy Krueger, MD, PhD; Micaela Rozenberg; Oded Volovelsky, MD, PhD

Sponsored by: Marinus Pharmaceuticals

WORKSHOP: Advocacy in the Education System at All Levels, Plum Blossom

Moderator: Kate Crossett

Speakers: Jennifer Flinn, EdD; Billy Holcombe, PhD; Shelly Meitzler; Nadia Vann, JD

DISCUSSION: TAND - Mental Health Issues in Living with TSC as an Independent and

Semi-Independent Adult, Edelweiss

Moderator: Derek Bauer, MD

Speakers: Jamie Capal, MD; Petrus de Vries, MBChB, FRCPsych, PhD

Sponsored by: TacomaBeast

Saturday (cont.)

2:30 p.m.-3:00 p.m. BREAK

Sponsored by: LivaNova

3:00 p.m.-4:30 p.m. Sixth Break Out Session

*PANEL: Finding your Way Home - Creative Housing Options/ Microboards, Grand Ballroom A

Moderator: Kelly Piacenti

Panelists: Deanna Eble, Esq.; John Hulbert, MD; Lisa Szilagyi

SPECIALTY: Managing Progressive Kidney Disease, Grand Ballroom B

Moderator: John Bissler, MD

Speakers: Chris Kingswood, MD; Joshua Samuels, MD, MPH

LECTURE: Gene Therapy – Hopes and Hurdles, Grand Ballroom D & E

Moderator: Michael Wong, MD, PhD

Speakers: Julian Martinez-Agosto, MD; Mustafa Sahin, MD, PhD; Kristi Viles, PhD, PMP

Sponsored by: BridgeBio

WORKSHOP: TAND - Feeding, Elopement and Potty Training, Plum Blossom

Moderator: Nathan Call, PhD, BCBA-D

Speakers: Joanna Lomas-Mevers, PhD, BCBA; Mindy Scheithauer, PhD, BCBA-D; Valerie

Volkert, PhD, BCBA-D

DISCUSSION: What's Next? Tools to Empower You on Your Medical Journey to an Adult

Neurologist, Edelweiss

Moderator: Klaus Werner, MD, PhD

Speakers: Derek Bauer, MD; Elizabeth Thiele, MD, PhD

4:30 p.m.–4:45 p.m. **BREAK**

4:45 p.m.-5:30 p.m. GENERAL QUESTIONS AND ANSWERS (Panel of Presenters), Grand Ballroom D/E

Moderator: Steve Roberds, PhD

Panelists: Derek Bauer, MD; John Bissler, MD; Petrus de Vries, MBChB, FRCPsych, PhD;

David Dunn, MD; Gerald Grant, MD; Lisa Henske, MD; Shafali Jeste, MD; Darcy Krueger, MD, PhD;

Angela Peron, MD; Elizabeth Thiele, MD, PhD

6:30 p.m.–8:30 p.m. Blue Night Dinner, Chantilly Ballroom

Host: Kari Luther Rosbeck Sponsored by: UCB, Inc.

Entertainment sponsored by: TSC Alliance Staff

8:30 p.m.–11:00 p.m. Parents and Dependent Adults Social Gathering, Stemmons

8:30 p.m.–11:00 p.m. **Teen Lounge Open**, *Obelisk A & B*

8:30 p.m. Adults with TSC Evening Activity, Plum Blossom

Event Leaders: Rob Grandia, Iris Mustich

Adults with TSC Evening Activity: Board Game Café

9:00 p.m. TSC Mom's Outing, Gossip Bar

Event Leader: Carly Kaye

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Sunday, July 31, 2022

7:00 a.m.-8:30 a.m. Breakfast, Chantilly Ballroom

7:30 a.m.–12:30 p.m. Day Camp, Atrium I – Check-In: Stemmons Ballroom 8:00 a.m.–12:00 p.m. TSC Store Open, in Conference Registration Area

8:30 a.m.-10:00 a.m. Seventh Break Out Session

PANEL: Other Organ Manifestations - Liver, Pancreas, Heart and Eyes, Grand Ballroom A

Moderator: Rajsekar Rajaraman, MD

Panelists: Mary Aronow, MD; Darcy Krueger, MD, PhD; David Rodriguez-Buritica, MD

SPECIALTY: Transition into Adulthood – Employment and Higher Education, Grand Ballroom B

Moderator: Julie Scroggins

Speakers: Deanna Eble, Esq.; Preston Fitzgerald; Jonathan Goldstein; Celie Johnson;

Kirk McCreary; Emily Racenet

LECTURE: TAND - Behavioral Interventions and Overview of Medications, Grand Ballroom D & E

Moderator: Billy Holcombe, PhD

Speakers: Petrus de Vries, MBChB, FRCPsych, PhD; David Dunn, MD; Tanjala Gipson, MD;

Shafali Jeste, MD

*WORKSHOP: Navigating Health Care System to Better Access Medication, Plum Blossom

Moderator: Mary Kay Koenig, MD

Speakers: Wynne Bird BSN, RN, CPN, CNRN; Jessica Krefting, BSN, RN; Ashley Pounders,

MSN, FNP-C

Sponsored by: Longboard Pharmaceuticals

DISCUSSION: Sexual Development for Dependent and Semi-Independent TSC Adults,

Edelweiss

Moderator: Shelly Meitzler Speaker: Jamie Capal, MD

10:00 a.m.-10:30 a.m. BREAK

10:30 a.m.-12:00 p.m. CLOSING SESSION: Driving the Future of TSC - Clinical Trials and Clinical Care,

Grand Ballroom D & E

Moderator: Anna Jansen, MD, PhD

Family Speakers: Dave and Rachel Johnson

Speakers: Martina Bebin, MD, MPA; Darcy Krueger, MD, PhD; Joel Moss, MD, PhD; Mustafa

Sahin, MD, PhD

Closing Remarks: Kari Luther Rosbeck

Sponsored by: Upsher-Smith Laboratories, LLC

Additional Support by: Lundbeck

^{*}All are welcome; however, these sessions may be most relevant to individuals living in the United States.

Conference Speakers



Ary Agami, President, TSC Alliance of Mexico, Cancun, Mexico

Ary Agami was born in Mexico in 1969 and is married with three girls. His second daughter, Larissa, is affected by TSC. Ary is an economist at the Tec de Monterrey and has run his own business for 22 years in the hospitality sector for interior design.



Dean J. Aguiar, PhD, Vice President, Translational Research, TSC Alliance, Spring Hill, TN

Dean joined the TSC Alliance in November 2018 with more than 17 years of research and development (R&D) leadership in biopharmaceutical and medical device industries, leading teams and technologies from discovery to investigational new drug and investigational device exemption, a pre-requisite for clinical trial evaluation. He brings an entrepreneurial and collaborative approach to R&D, identifying opportunity, defining strategy and developing a scientific data package to warrant clinical translation of technologies to benefit the patient, the family and care provider. Dean has a breadth of pharmaceutical R&D experiences from early discovery to clinical translation that provides a unique perspective on project management and mitigating risk. Dean's early career landed him at Pfizer, where he led cross-functional project teams in the areas of inflammation spanning discovery to preclinical development of both small molecules and protein biologic drugs. He played a key role while ensuring a robust decision funnel for translatable cell, animal models and biomarkers of human disease. Dean earned his PhD in Biochemistry from Rush University at Rush Presbyterian St. Luke's Medical Center in 1996 and completed post-doctoral training at the University of Minnesota.



Mary E. Aronow, MD, Assistant Professor of Ophthalmology, Massachusetts Eye and Ear, Harvard Medical School, Boston, MA

Dr. Aronow is an ocular oncologist who specializes in the full spectrum of adult and pediatric eye tumors. She received her medical degree from Yale University School of Medicine and completed her ophthalmology residency training at the Cleveland Clinic. She did fellowship training in ophthalmic oncology at the Cleveland Clinic and completed a second fellowship in medical retina at the National Eye Institute/National Institutes of Health. Dr. Aronow served on the faculty at the Johns Hopkins Wilmer Eye Institute for several years and built a full-service eye cancer service before being recruited to the faculty at Harvard as an Assistant Professor of Ophthalmology in 2017. She has served on the TSC Alliance's Professional Advisory Board since 2016.



Derek Bauer, MD Assistant Professor, University of Virginia Neurology, Charlottesville, VA

Dr. Bauer is an adult epileptologist at the University of Virginia, where he co-directs the TSC Clinic. He is also an adult with TSC and was diagnosed at age 12. He is a member of the TSC Alliance Board of Directors.



Martina Bebin, MD, MPA, Professor of Neurology and Pediatrics, University of Alabama at Birmingham

Martina Bebin MD, MPA, is Professor of Neurology and Pediatrics at the University of Alabama at Birmingham. Her primary research interest is in tuberous sclerosis complex (TSC); she serves as co-director of the UAB TSC Clinic, which is a Center of Excellence and offers comprehensive multidisciplinary TSC care for all ages. She has been involved in clinical epilepsy and TSC research for more than 20 years. Currently, she is the Principal Investigator (PI) for the NIH-funded PREVeNT Trial, the first epilepsy prevention trial for infants with TSC. Also, she is an original member of the TSC Clinical Research Consortium and a site PI for several NIH-funded TSC clinical research efforts over the past 10 years.

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Andrea Beebe, TSC Parent and Advocate, Clayton, DE

Andrea is mom to a 3-year-old boy named Parker diagnosed with TSC. Andrea is a special education early childhood teacher in the local school district. The family learned of TSC and Parker's complications 5 weeks before his birth. Following Parker's birth, he required immediate heart surgery and is followed by numerous doctors at Children's Hospital of Philadelphia and Al Dupont Children's Hospital in Delaware. He is currently in preschool and receives speech and language, occupational, and physical therapies. He is an energetic, happy, and loveable young boy with an amazing family and support system. The Beebe family made it their mission to spread awareness about TSC following Parker's diagnosis. Through multiple fundraising campaigns, including TSC Awareness Jeans days at Andrea's work and a very successful "Hot Ones"-style virtual wing eating competitions, the Beebe family, friends and support system have raised more than \$150,000 in the last three years.



Wynn Bird, BSN, RN, CHRN, CPN, Nurse Care Coordinator, Children's Hospital Colorado, Aurora, CO

Wynne Bird, BSN, RN, CNRN, CPN began her nursing career after graduating with her BSN from the University of Northern Colorado. She has worked as the Tuberous Sclerosis and Epilepsy Surgery Nurse Care Coordinator at Children's Hospital Colorado for the past 11 years. Previously, she worked in the neuroscience inpatient unit for more than 20 years. Her expertise and passion have focused on improving family-centered care and increasing education for specialized nursing



John J. Bissler, MD, Federal Express Chair of Excellence, Professor of Pediatrics, Director, Tuberous Sclerosis Center of Excellence, Director, Division of Nephrology at St. Jude Children's Research Hospital, Director, Division of Nephrology at Le Bonheur Children's Hospital, Memphis, TN

Professor John Bissler saw his first patient with TSC more than 35 years ago. Professor Bissler was struck by the lack of accurate information and treatment options for TSC patients at that time and has focused his career on the care of TSC patients with renal disease. Professor Bissler's work in this area has ranged from developing the steroid protocol to greatly reduce the pain of the angiomyolipoma embolization procedure to being the principal investigator for the use of mTORC1 inhibitors for TSC renal disease. Professor Bissler has seen more than 1,500 individual TSC patients in the United States and many more internationally. Professor Bissler continues to focus his research on TSC renal disease and is currently looking for therapies for the cystic disease, which still has no approved therapies. Professor Bissler maintains a busy clinical practice as the Director of the TSC Center of Excellence in Memphis, TN, one of the largest TSC Centers in the USA. Professor Bissler also specializes in the renal care of the child with cancer and is the Director of Nephrology at St. Jude Children's Research Hospital and Le Bonheur Children's Hospital, both in Memphis.



Jeffrey P. Blount, MD, MPH, Division Director, Division of Pediatric Neurosurgery, University of Alabama at Birmingham, AL

Jeffrey P. Blount is the Raymond J. Harbert Professor and Chair of Pediatric Neurosurgery at the University of Alabama at Birmingham. He is a graduate of University of Rochester School of Medicine, the University of Minnesota Neurosurgery Residency program and the fellowship in Pediatric Neurosurgery at the Hospital for Sick Children. His primary academic interests center on surgical treatment of epilepsy in children and management of Spina Bifida in children and adults.



Bridget Britt, TSC Parent, TSC Alliance Volunteer, San Leandro, CA

Bridget is a mom, an elementary school teacher and an advocate for the TSC community. Bridget has two daughters, one of whom was diagnosed with TSC. Since her daughter Molly's diagnosis, Bridget has served as a Step Forward to Cure TSC Walk Chair, advocated for funding for TSC during the March on Capitol Hill and connected with other TSC families throughout their TSC journeys.



Nathan Call, MD, Professor of Pediatrics in the Emory University School of Medicine, Atlanta, GA

Dr. Call is a Professor of Pediatrics and Associate Chief of the Division of Autism & Related Disorders in the Emory University School of Medicine. He has worked at Marcus Autism Center since 2006, where he has held many positions as a clinician and leader of clinical programs. He currently serves as the Director of Clinical Affairs & Interim Vice President for the center. In this role he oversees all clinical activities and works with providers and other center leaders to develop new clinical innovations. Dr. Call has an active research agenda and has published over sixty papers and book chapters on the treatment of challenging behavior exhibited by individuals with autism and related disorders.



Jamie K. Capal, MD, Associate Professor of Pediatrics and Neurology, University of North Carolina at Chapel Hill and Carolina Institute for Developmental Disabilities, Chapel Hill, NC

Dr. Capal is an Associate Professor of Pediatrics and Neurology. She is board-certified in Pediatrics, Neurology with special qualification in child neurology, and Neurodevelopmental Disabilities. She received her training at Cincinnati Hospital Medical Center and stayed on as faculty there until the spring of 2020. While at Cincinnati Children's Hospital, she was a member of the multi-disciplinary Tuberous Sclerosis Complex Clinic and was involved in several research studies funded by the National Institutes of Health and TSC Clinical Research Consortium (TACERN), including determining biomarkers for autism spectrum disorder (ASD) in young children with TSC, ages birth to 36 months; determining the timing and relationship between seizures, cognition and development of ASD; evaluating the broader autism phenotype in ASD; and examining common features of ASD among different single-gene disorders with high prevalence of ASD (TSC, Phelan-McDermid syndrome, PTEN Hamartoma syndrome). She has since joined the University of North Carolina at Chapel Hill where she has a joint appointment in neurology and the Carolina Institute for Developmental Disabilities. Her clinical and research interests focus on children with neurodevelopmental and neurogenetic conditions, including autism spectrum disorders, TSC and Angelman syndrome. She has transitioned to UNC Chapel Hill with a vision to improve care for individuals with TSC. She also started a TAND clinic.



Lisa D. Carlton, Vice President, Regulatory Affairs, REGENXBIO Inc., Rockville, MD

Lisa is currently Vice President of Global Regulatory Affairs at REGENXBIO Inc. She has more than 25 years of experience in regulatory affairs and medical/technical writing in the government/nonprofit sectors, with the majority of her career spent in the pharmaceutical/biotech industry. After completing her BS in Biochemistry at North Carolina State University, Lisa obtained an MS degree in Medicinal Chemistry and PhD in Pharmaceutics from the University of North Carolina in Chapel Hill followed by postdoctoral research at NIH's National Institute of Allergy and Infectious Disease in the Laboratory of Immunogenetics.



Jocelyn Cenna, TSC Clinic Ambassador, TSC Center of Excellence, University of Maryland Medical Center, Baltimore, MD

Jocelyn became a caretaker for her adult brother Frank who has TSC when their sister passed in 2011 and Jocelyn moved him to Maryland. Frank was not diagnosed with TSC until one year following brain surgery in 1976 when he had to drop out of college. While Jocelyn was working in Washington, DC in the late 1990s, she noticed a TSC medical conference was being held and stopped by to pick up brochures to learn more about her brother's rare disease. Over the past 20 years Jocelyn participated in many Step Forward to Cure TSC walks in Silver Spring, MD and Washington, DC; attended DC fundraising events; Maryland Awareness and Advocacy events in Annapolis, MD; and the Virtual March on Capitol Hill efforts to secure federal funding for TSCRP. During a volunteer event Jocelyn met a distant cousin, Sean Shillinger and his wife Lauren, whose daughter also has TSC. She became a TSC Clinic Ambassador for Johns Hopkins Kennedy Krieger Institute in 2014, a TSC Ambassador for Inspire collaboration website in 2019 and the TSC Clinic Ambassador for the TSC Center of Excellence at the University of Maryland Medical Center in 2020. Frank and Jocelyn have attended prior World TSC Conferences in DC in 2014 and in Dallas in 2018. Jocelyn has two BS degrees from Carnegie Mellon University, an MBA from Marymount University and an Alumnae of the Year for University of California Irvine Division of Continuing Education. She is a retired certified Project Management Institute Project Management Professional. After working for General Motors, she moved into the defense world and was in charge of various military defense programs and led a group of Program/Project Managers across the U.S.

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Sara Chieffo, Vice President, Government Affairs, League of Conservation Voters, Washington, DC

Sara Chieffo joined the Board of Directors of the TSC Alliance in 2017 and currently serves as chair of the Government Relations Committee. Sara first got involved with the TSC Alliance shortly after her oldest daughter was diagnosed with TSC in 2013 and her first action was to participate in the March on Capitol Hill. Sara is extremely proud of the incredible work of the Government Action Team to build a grassroots advocacy powerhouse that has raised tens of millions of federal dollars into research and a cure for TSC and the resulting breakthroughs are already making a difference in her daughter's life. In her professional life, Sara serves as the League of Conservation Voters' Vice President of Government Affairs where she works to advance LCV's federal legislative and administrative goals with a focus on climate change and energy policy. Her work spans legislative strategy, executive branch advocacy, coalition-building, and communications. Sara's background also includes non-profit fundraising, state legislative work, and electoral campaigns. Sara holds a master's degree from Carnegie Mellon University, a B.A. from Georgetown University, and is a graduate of the Coro Fellowship in Public Affairs in Pittsburgh, PA.



Asim F. Choudhri, MD, Professor of Radiology, Ophthalmology and Neurosurgery, University of Tennessee Health Science Center, and Chief of Neuroradiology and Director of MRI at Le Bonheur Children's Hospital, Memphis, TN

Dr. Choudhri is Professor of Radiology, Ophthalmology, and Neurosurgery at the University of Tennessee Health Science Center, and Chief of Neuroradiology and Director of MRI at Le Bonheur Children's Hospital in Memphis, TN. He has an interest in neurocutaneous syndromes, brain tumors, and epilepsy.



Danielle G. Clark, MPH, MS, RD, TSC Alliance Adult Regional Coordinator, San Antonio, TX

Danielle Garcia Clark has been a Registered Dietitian for more than 15 years and is currently working as a Renal Dietitian. Danielle has been an active volunteer with the TSC Alliance since 2018, serving as an Adult Regional Coordinator and Social Media Chair for the adult community. Danielle has participated in the March on Capitol Hill since 2019 and has been a peer reviewer for the Congressionally Directed Medical Program with the Department of Defense. In her years as a Registered Dietitian, listening to patients and their needs have helped her develop a good rapport with others. Her interpersonal experiences have not only assisted her with patients but also herself through her TSC journey in creating healthy coping skills.



Rohini Coorg, MD, Assistant Professor, Baylor College of Medicine/Texas Children's Hospital, Waco, TX

Dr. Rohini Coorg is a board-certified child neurologist, neurophysiologist and epileptologist with a special interest in the medical and surgical management of epilepsy in tuberous sclerosis complex at Texas Children's Hospital (TCH). Her roles at TCH include serving as the clinic director of a multidisciplinary clinic for TSC patients, which she helped formally establish in 2015. Dr. Coorg is also medical director of the epilepsy monitoring unit (EMU) where she interprets diagnostic and pre-surgical continuous EEG-video studies and, with a whole team at TCH, is an active participant in the care of children undergoing epilepsy surgery. Dr. Coorg completed her neurophysiology and epilepsy training at Washington University in St. Louis/St. Louis Children's Hospital and child neurology training at University of California, Irvine and has practiced at Texas Children's Hospital since 2014.



Peter B. Crino MD, PhD, Professor and Chair, Department of Neurology, University of Maryland School of Medicine, and Chair, TSC Alliance Board of Directors, Baltimore, MD

Peter B. Crino is Professor and Chair of Neurology at the University of Maryland, Baltimore. He received his BA from Binghamton University, MD from Yale University and PhD in neuroscience from Boston University. He completed a medical internship at Yale-New Haven Hospital and then a neurology residency at the University of Pennsylvania. He was selected as a Howard Hughes Medical Institute Physician Post-Doctoral Fellow in Molecular Neuroscience at the University of Pennsylvania with clinical training in epilepsy. Dr. Crino has received the Esther A. and Joseph Klingenstein Award in Neuroscience (1999), a Lifetime Service Award from the TSC Alliance (2008), the University of Pennsylvania Leonard Berwick Memorial Excellence in Teaching Award (2010), the Fritz Dreifuss Award for Translational Research in Epilepsy from the AES (2016) and the Jacob Javits Neuroscience Research Award in 2021 from the NINDS. Dr. Crino was awarded the inaugural Dr. Richard and Kathryn Taylor Endowed Professorship in Neurology at the University of Maryland School of Medicine. In 2019, Dr. Crino received funding from the state of Maryland to support the new Clinical Center for Adults with Neuro-Developmental Disabilities (CCAND) at University of Maryland Medical Center and University of Maryland School of Medicine. This program provides comprehensive care for adults with epilepsy and intellectual disability. Dr. Crino also directs the TSC Center of Excellence in Maryland. Dr. Crino's laboratory has maintained a continuously NINDS-funded translational research program studying mechanisms of altered brain development associated with epilepsy, intellectual disability, and autism for the past 22 years. He has more than 170 published papers, reviews and book chapters and numerous international presentations.



Kate Barbato Crossett, Med, TSC Alliance Adult Regional Coordinator, Education Parent Mentor and Special Education Administrator, Philadelphia, PA

Kate was diagnosed with tuberous sclerosis complex as a toddler in 1986 at Milton Hershey Medical Center. After some turbulent times managing Kate's seizures and grappling with her diagnosis, Kate's parents-Steve & Abbe-joined the efforts as volunteers of the original National Tuberous Sclerosis Association for many years. Kate graduated from the University of Pittsburgh in 2007, and she joined Teach For America as a Corps Member in Philadelphia teaching Special Education Math and Science. During this time, she earned her Master's in Education and certification in Special Education at Chestnut Hill College. For a brief time following her first few years of teaching, Kate ventured to Florence, Italy earning a Post-Baccalaureate degree in Art Restoration and Conservation. She returned to the United States and to a career in education in 2011. Kate earned her Principal Certification from Relay Graduate School of Education in New York City in 2016, and she has been a Special Education Administrator in Philadelphia for about 10 years now, originally serving as a founding Assistant Principal at a middle school in North Philadelphia. Kate resumed her volunteer efforts with the TSC Alliance in 2019, working closely with the Adult Regional Coordinators and Education Parent Mentors, raising funds for and participating in Step Forward to Cure TSC and advocating at the March on Capitol Hill for several years now. Kate is so incredibly thankful for the extended family she has found here and to be participating in the World TSC Conference this year with the support of her husband–Rob, son–Legend, and her parents.



Daniel J. Curry, MD, Texas Children's Hospital, Houston, TX

Daniel Curry, MD, is The John S. Dunn Foundation Endowed Chair for Minimally Invasive Epilepsy Surgery at Texas Children's Hospital and a Professor of Pediatric Neurosurgery at Baylor College of Medicine. His clinical interests include minimally invasive treatment in epilepsy surgery, treatment of pediatric movement disorders and chronic pain. Dr. Curry's research interests include study of the mechanisms of dystonia and non-invasive detection of epilepsy networks.



Peter E. Davis, MD, Boston Children's Hospital Department of Neurology, Boston, MA

Dr. Davis is a pediatric neurologist at Boston Children's Hospital. His research uses clinical data and computational EEG methods to identify risk factors for epilepsy and neurodevelopmental disorders in children with TSC. Clinically, Dr. Davis sees children with autism, intellectual disabilities, epilepsy and other neurodevelopmental disorders in the Multi-disciplinary Tuberous Sclerosis Clinic and Autism Spectrum Center at Boston Children's Hospital.

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Prof. Petrus J. de Vries, MBChB, FRCPsych, PhD, Sue Struengmann Professor of Child & Adolescent Psychiatry, University of Cape Town, South Africa

Petrus de Vries is the Sue Struengmann Professor of Child & Adolescent Psychiatry and Founding Director of the Centre for Autism Research in Africa at the University of Cape Town. He trained in Medicine at Stellenbosch University in South Africa before moving to the UK where he completed his clinical training in Psychiatry and Child & Adolescent Psychiatry and a PhD in Developmental Neuroscience at the University of Cambridge. He returned to South Africa in 2012. Apart from his interest in autism, he has a longstanding clinical research programme in TSC and its associated neuropsychiatric disorders (TAND). He was the chairman of the International Neuropsychiatry Panel that coined the term "TAND" and was lead developer and author of the TAND Checklist. He now leads the TANDem project with Prof. Anna Jansen. Prof. de Vries has been actively involved with many TSC user/caregiver organizations over the last 25 years, including the TSA UK, the TSC Alliance and many TSCi organizations around the globe. He was awarded the Manuel R. Gomez Award (2014) and the Bourneville Prize (2021) for his contributions to TSC and TAND.



David W. Dunn, MD, Professor of Psychiatry and Neurology, Indiana University School of Medicine, Indianapolis, IN

David W. Dunn, MD, is the Arthur B. Richter Professor of Child Psychiatry and Professor of Psychiatry and Neurology at Indiana University School of Medicine. He is residency training director for the child and adolescent psychiatry program and the combined pediatrics-psychiatry-child and adolescent psychiatry (triple board) program. Dr. Dunn is certified by the American Board of Psychiatry and Neurology in Neurology with Special Competence in Child Neurology, Psychiatry, Child and Adolescent Psychiatry, and Epilepsy. He is a member of the Scientific Advisory Board of the TSC Alliance and has served as chair of the Department of Defense Tuberous Sclerosis Complex Research Program.



Deanna M. Eble, Esq, Partner, Russo Law Group, Garden City, Islandia, Lido Beach and Manhattan, NY

Deanna M. Eble is a Partner at Russo Law Group, P.C. She focuses on Special Needs Planning, Elder Law, Estate Planning and Medicaid Planning and has practiced in these areas for 20 years. Deanna is a member of the New York State Bar Association, Nassau County Bar Association, Nassau County Women's Bar Association, Elder Counsel and is admitted to the New York Bar. She earned her law degree at Hofstra University School of Law. Deanna frequently presents seminars on Special Needs and Estate Planning throughout Nassau and Suffolk counties. As an attorney, Deanna has helped hundreds of families with all levels of planning. She assists families who have a child or loved one born with special needs and as a disability lawyer in New York. Deanna works as an advocate for their loved one throughout his or her life from on-going decision making (guardianship), applying for Supplemental Security Income and New York Medicaid and setting up special needs trusts.



Preston Fitzgerald, Former TSC Alliance Future Leader, Sachse, TX

Preston was born in 1999 in the Dallas/Fort Worth area. He was diagnosed with TSC when he was six months old. Preston has tumors in his brain and both kidneys. The brain tumor causes him to have occasional partial seizures. Preston and his family have been advocates for the TSC family ever since he was diagnosed. He was a TSC Alliance Future Leader in 2020-2021 and is currently a Mentor for the current class. He graduated from the University of North Texas in May of 2022 with a degree in hospitality. In the future, Preston hopes to continue his advocacy efforts while using his college degree to travel the world.



Carla Fladrowski, Co-Chair TSC Italy, Manager International Relations, Co-Chair European Tuberous Sclerosis Complex Association (E-TSC), Member of Tuberous Sclerosis Complex international (TSCi) Working Group, Monza, Italy

Carla has two sons Leo, 27 and Jake, 24. Her introduction to TSC was in 1997 after Jake was diagnosed in utero. Since her journey began, she has battled for gold standards in care, dignity and respect in treatment, further research, empowerment and awareness on TSC. Tuberous Sclerosis Complex Italy (Associazione Sclerosi Tuberosa aps) was founded in May 1997 and this year goes "Silver." Carla has been responsible within TSC Italy for networking between the different TSC communities and key clinicians around the world since 2009 and in 2021 was elected Co-Chair. Carla was also involved in re-establishing the European Tuberous Sclerosis Complex Association (ETSC), which this year celebrates a decade of #makingTSCmatter. She has also been a member of the Tuberous Sclerosis Complex International Working Group since 2011. Carla co-founded the ETSC DEA Project (Disseminating Expertise and Achievement), which aims to establish a holistic approach in the treatment of TSC, promote multi-disciplinary working and create a readily accessible, active and sustainable network within and between the EU and internationally.



Jennifer Flinn, EdD, TSC Canada Board of Directors, Ottawa, ON, Canada

Jennifer Flinn is a Vice-Principal at a large kindergarten to grade 6 elementary school at the Ottawa Catholic School Board in Canada. She taught in a classroom for 15 years before moving into various board level positions. Jennifer completed undergraduate studies at the University of Ottawa in psychology and education. She earned a Master's of Education at the University of Western Ontario, with thesis work on the educational impacts of tuberous sclerosis complex. Jennifer then completed her doctorate in Educational Leadership at UWO, focusing on the intersection of rare chronic health conditions, policy, leadership and inclusion in education systems. She has sat on the board of directors of TSC Canada since 2008 and has been a member of TSCi since 2009. Jennifer is a member of the The TAND Consortium and leads the Scholastic Cluster as part of TANDem, an international multi-disciplinary mobile-health project with the purpose of empowering families and individuals around the world who live with TSC. She has authored a children's book called "Princess Katie the Brave: A Story About Living with TSC" to help young children better understand the condition.



Sarah Friebert, MD, Director, Pediatric Palliative Care, Akron Children's Hospital, Akron, OH

Dr. Sarah Friebert is founder and director of the Haslinger Family Pediatric Palliative Care Center as well as the Emily Cooper Welty Expressive Therapy Center at Akron Children's Hospital. Dr. Friebert completed residency in pediatrics at The Children's Hospital of Philadelphia and a fellowship in pediatric hematology/ oncology at Rainbow Babies and Children's Hospital. Under her direction, the Haslinger Center features an accredited palliative care fellowship program. The team she leads offers physical, psychosocial, emotional and spiritual support as they serve children with serious conditions and their families in inpatient, outpatient and home and community settings. In addition to clinical care, the Center is active in education, outreach, advocacy and research. She is Professor of Pediatrics at Northeast Ohio Medical University, where she is a founding member of the Office of Palliative Care and active in medical student education. Named as a Visionary in Hospice and Palliative Medicine by the American Academy of Hospice and Palliative Medicine (AAHPM) and recipient of the Palliative Medicine Community Leadership Award from the Project on Death in America and AAHPM, she is the author of multiple articles and book chapters and an active research scientist, serving as program director for palliative care research. She was co-principal investigator for a Centers for Medicare & Medicaid Health Care Innovation Award examining care coordination for children with complex medical conditions. Currently, Dr. Friebert is principal investigator on multiple palliative care-related research initiatives, including a multi-site study on adolescent advance care planning. Outside of Akron Children's Hospital, she is active in multiple local, regional, state and national educational, philanthropic and palliative care organizations. When not engaged in serving the palliative care community, she can be found in her kayak.

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Lara A. Friel, MD, PhD, Associate Professor, University of Texas Health Science Center at Houston/ McGovern Medical School, Houston, TX

Lara A. Friel, MD, PhD, is a Maternal Fetal Medicine specialist at UTHealth, the University of Texas Health Science Center at Houston/The McGovern Medical School. She is an Associate Professor in the Department of Obstetrics, Gynecology and Reproductive Sciences. Dr. Friel's research interests include genetics and pregnancy complications; she teaches and mentors Maternal Fetal Medicine fellows, Obstetrics and Gynecology residents, UTHealth medical students and genetic counseling students. She also supervises the research mission of the 12-member genetic counseling team at UTHealth. Frequently, she presents the genetics update for practicing obstetricians at the Clinical and Scientific Meeting of the American College of Obstetrics and Gynecology; authors the chapter, "Pregnancy Complicated by Disease," for both the Merck Manuals home and professional editions; and serves on the Society for Maternal Fetal Medicine's patient education committee. She has been the UTHealth principal investigator for several multicenter studies, including projects in preterm premature rupture of membranes, normal blood pressure in pregnancy and preeclampsia.



Michael Frost, MD, St. Paul, MN

Michael Frost, MD, is board certified in Pediatrics, Child Neurology, Clinical Neurophysiology and Epilepsy and has served as the principal investigator for numerous clinical drug trials with a particular interest in the treatment of TSC in the epilepsy population. Before his retirement in 2021, Dr. Frost was the Director of Pediatric Service for the Minnesota Epilepsy Group, PA®; Associate Medical Director for Neurosciences and Medical Director of Epilepsy and Clinical Neurophysiology at Children's Hospitals and Clinics-Minnesota in St. Paul; and Clinical Professor in the Department of Neurology at the University of Minnesota in Minneapolis. Dr. Frost, along with Dr. John Hulbert, founded the Tuberous Sclerosis Complex Clinic Without Walls in the Upper Midwest and continues to serve on the TSC Alliance's Professional Advisory Board.



Zoë Fuchs, Science Project Manager, TSC Alliance, San Francisco, CA

Zoë joined the TSC Alliance in March 2019 after volunteering with the organization in early 2018. She graduated from the University of California, Santa Barbara with a degree in Chemical Engineering. Zoë has a passion for preclinical and translational research. As an undergraduate, she studied methods to improve chemotherapeutic efficacy via liposomes and polymer-drug conjugates. Following graduation, she worked for UCLA's Translational Oncology Research Laboratory, where she studied leukemia and lymphoma and tested novel treatments in a high-throughput lab. She also worked as a Medical Scribe for George Washington (GW) University in a wide range of specialist clinics, spending most of her time working at the GW Cancer Center.



Dr. Pradnya Gadgil, Director, TSC Clinic, Kokilaben Ambani Hospital, Mumbai, India

Dr. Pradnya Gadgil is a paediatric neurologist with a special interest in complex epilepsy. She anchors the paediatric epilepsy surgery program, a comprehensive program offering resective surgeries with 1 or 2 stage (SEEG) evaluations as needed as well as palliative procedures such as VNS, DBS etc. Dr. Gadgil works closely with the national epilepsy associations to raise epilepsy awareness, conduct rural epilepsy camps, etc. Her clinic was fortunate to be designated as the first TSC clinic in India. The clinic supports the TSC Alliance of India, which though quite young, has been proactive in advocacy and raising awareness for TSC. Dr. Gadgil's other passion is teaching. Apart from being a designated teacher for the local postgraduate paediatric and paediatric neurology programs, she has had the chance to be the country lead for India for the international paediatric epilepsy teaching collaboration with the British Paediatric Neurology Association.



Joao García, MD, Director, Neural Clinic, Mérida, México

Dr. Garcia graduated from the Autonomous University of Guadalajara and trained in pediatrics and pediatric neurology at the National Institute of Pediatrics of Mexico City. He is currently director of the Neural group and also of the TSC clinic in Mérida, Yucatán, the first clinic established in Mexico.



Tanjala T. Gipson, MD, Director, TSC-Associated Neuropsychiatric Disorders Unit, Le Bonheur Children's Hospital, Memphis, TN

Dr. Tanjala Gipson is both a child neurologist and neurodevelopmental disabilities specialist. She founded the TSC Center of Excellence at Kennedy Krieger Institute and established an interdisciplinary model of care for children and adults with TSC-associated neuropsychiatric disorders. She has now moved to LeBonheur Children's Hospital and the Boling Center for Developmental Disabilities at the University of Tennessee Health Sciences Center in Memphis, TN where she has established a clinical and research unit dedicated to TSC-associated neuropsychiatric disorders.



Jen Glass, CGC, Cincinnati Children's Hospital, Cincinnati, OH

Jen fell in love with genetics at the age of 14 at a summer academic camp. She got her Bachelor's degree in Genetics at Purdue University and her Master's in Genetic Counseling at Case Western Reserve University. Jen has been working at the TSC Center of Excellence at Cincinnati Children's Hospital since 2010, where she also manages the team of genetic counselors working in the Cincinnati Children's Hospital Genetics and Genomics Diagnostic Laboratory. In addition to providing genetic counseling to families with TSC, Jen also supervises students and is involved in TSC clinical research. Jen has a particular interest in patients with persistent negative genetic testing and developing pregnancy related recommendations for patients and/or pregnancies affected by TSC. Jen loves working with TSC patients and has been excited to become involved with the TSC Alliance by giving talks and serving on the TSC Alliance Professional Advisory Board.



Jonathan Goldstein, Former TSC Alliance Future Leader, New York, NY

Jonathan is a TSC patient who is focused on promoting health equity and increasing access to affordable, quality care for all individuals--especially those who have been historically disenfranchised in our health systems. He is a management professional with experience in business unit strategy planning and development, marketing, operating model design and implementation. Jonathan has led initiatives centered on behavioral health, Medicare, labor and public policy analysis, digital strategy frameworks, payer agnostic product solutions, and has experience leading teams, spearheading diversity, equity and inclusion initiatives and working in cross-functional, matrixed organizations. Jonathan is based in New York City and currently serves on the TSC Alliance Equity, Diversity, & Inclusion Taskforce and on the TSC Alliance Board of Directors Outreach Committee. He previously participated in the TSC Alliance Future Leaders Program and enjoys mentoring young adults with TSC.



Elizabeth S. Gosnell, DMD, MS, Associate Professor-Clinical, Cincinnati Children's Hospital Medical Center, Cincinnati, OH

Elizabeth S. Gosnell, DMD, MS, is a pediatric dentist and associate professor at Cincinnati Children's Hospital Medical Center (CCHMC) in Cincinnati, Ohio. She is a full-time attending physician, educates residents, maintains a clinical practice and conducts clinical research. She specializes in infant oral healthcare and caring for children, adolescents and adults with special healthcare needs. Locally, she serves as a member of the craniofacial anomalies team and as a consultant and providing dentist for the TSC team at CCHMC. Nationally, she is on the qualifying exam committee for the American Board of Pediatric Dentistry. She serves on the Professional Advisory Board for the TSC Alliance and has assisted in updating oral health recommendations for the TSC consensus statements published in 2012 and 2021. Dr. Gosnell is the chair of the Council on Clinical Affairs, committee on special health care needs in the American Academy of Pediatric Dentistry. Her research focuses on behavior guidance techniques and helping children manage anxiety in the dental clinic. Also, her research focuses on the oral health conditions and challenges in receiving oral healthcare for children with special healthcare needs.

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Althea Grace, Musician, TSC Parent, Los Angeles, CA

Fresh off a powerful run and Top 40 placement on ABC's American Idol, 21-year-old Althea Grace and her story touched the hearts of millions. Althea's first love was music, and she describes herself as, "Pop, but not modern-pop, more like The Faces or The Beatles type of pop." She was featured on the Los Lobos version of the Shins' song "The Fear." She was also previously credited for writing on Bramhall's album "Shades," including the track "Everything You Need" featuring Eric Clapton. Althea she was honored in 2021 at Comedy for a Cure 19.5 with the TSC Champion Award and performed at the 20th Anniversary Comedy for a Cure.



Rob Grandia, TSC Alliance Board of Directors, Adult Regional Coordinator, Riverside, CA

Rob Grandia is an adult with TSC and has served as a volunteer for the TSC Alliance for many years, moderating Regional TSC Conferences, Marching on Capitol Hill, serving on panels at the 2014 and 2018 World TSC Conferences and participating in the Outreach Committee. He currently serves as a TSC Alliance Adult Regional Coordinator. He has three children with TSC.



Rylee Grandia, TSC Alliance Volunteer and Former Future Leader, Riverside, CA

Rylee Grandia is a young adult with TSC who was diagnosed at 18 months old. Rylee's father and two younger brothers also have TSC, each with different manifestations and levels of severity. Over the years Rylee has helped her parents in a caretaking role of her brothers learning to manage behaviors, assisting with schoolwork and medication. She has also volunteered with the TSC Alliance at Step Forward to Cure TSC walks, Comedy for a Cure and TSC Regional Conferences. In 2017 Rylee was a TSC Alliance Future Leader and participated in the March on Capitol Hill in 2017, 2018 and online in 2021 and 2022. She wrote a blog as a Future Leader titled "The Life of Ry" that discussed her TSC journey. Rylee attended the 2018 World TSC Conference, supporting young adults; she also wrote an article in the Winter 2018 Perspective magazine. The Grandia family was recognized at Comedy for a Cure in 2018 with the Courage in Leadership Award in which Rylee played a role in the family being nominated; she created artwork with her brothers that was auctioned off at the event. After high school, Rylee attended junior college with a focus on early childhood development, but with the COVID epidemic and need for instructional aids in the public school system, she decided to put college on hold. She is currently an instructional aid and has found her purpose in supporting students that struggle with academic obstacles. Growing up within the TSC community and seeing the support from the TSC Alliance has helped Rylee see the difference one person can make in the lives of others.



Shannon Grandia, TSC Alliance Adult Regional Coordinator, Riverside, CA

Shannon Grandia is a spouse and a mother of three diagnosed with TSC. She has been the family caretaker with each member in the family battling different manifestations and different levels of severity. Shannon has been a public education teacher for 20 years with a Master's degree in multi-cultural education and recently completed a reading certificate program from USC. She first became involved with the TSC Alliance by participating in the local Step Forward to Cure TSC in 2010. From there Shannon and her husband Rob became Adult Regional Coordinators in 2014, supporting adults in 12 states. She participates in Adult Topic Calls and has been a speaker at multiple TSC Alliance educational events including the 2014 and 2018 World Conferences and local regional conferences. Shannon has participated in March on Capitol Hill for the past eight years. In 2016, Shannon received the Volunteer of the Year Award, and in 2018 the Grandia family was recognized at Comedy for a Cure with the Courage in Leadership Award. Shannon is on the Comedy for a Cure and the Step Forward for a Cure planning committees, and she and her husband were the Southern California walk chairs in 2018 and co-chairs in 2019. In August 2020, Shannon was given the opportunity to be a speaker in a TSC Alliance webinar that focused on COVID-19 and Going Back to School. Over the years the TSC Alliance and TSC community have become family to Shannon, and she knows the importance of supporting those living with TSC in all stages of the journey as her children were diagnosed very young and are now 21, 18 and 15 years old.



Gerald Grant, MD, Professor and Chair of Neurosurgery, Duke University, Durham, NC

Dr. Gerald Grant is currently an Endowed Professor in the Department of Neurosurgery and Chief of Pediatric Neurosurgery at Stanford University. In April, Dr. Grant will be moving back to Duke to be Professor and Chair of the Department of Neurosurgery. Dr. Grant graduated with a Bachelor of Science degree in Neuroscience from Duke University and completed medical education at Stanford University School of Medicine. Dr. Grant completed residency training at the University of Washington and completed a fellowship in pediatric neurosurgery at Seattle Children's Hospital. Dr. Grant then served active duty in the United States Air Force and was deployed to Iraq in support of Operation Iraqi Freedom. Dr. Grant joined the faculty at Duke in 2006 and was recruited back to Stanford in 2013. Dr. Grant has written over 270 peer-reviewed manuscripts and over 40 book chapters. He is a Diplomat of the American Board of Neurological Surgery and is also board certified in Pediatric Neurosurgery by the American Board of Pediatric Neurological Surgery. Dr. Grant is a fellow of the American College of Surgeons and the American Association of Neurological Surgeons. He has a specialized clinical interest in the surgical management of epilepsy in children.



Nishant Gupta, MD, Scientific Director, The LAM Foundation, Cincinnati, OH

Dr. Nishant Gupta is an Associate Professor at the University of Cincinnati College of Medicine where he also serves as the Director of the Interstitial and Rare Lung Disease program. He is also the Scientific Director of The LAM Foundation and the medical director of the LAM clinic network. Dr. Gupta specializes in interstitial lung diseases, and his work is focused on defining the natural history, improving detection and developing targeted therapies for patients with rare lung diseases such as LAM.



Latrice Hamilton, Licensed Professional Counselor Supervisor, TSC Alliance Volunteer, Memphis, TN

Latrice Hamilton is a wife to an amazing husband, Terrence Hamilton, and mother to one marvelous son, Terrence Hamilton Jr. (16). She works as a Licensed Professional Counselor Supervisor where she specializes in treating individuals of all ages with autism, ADHD, anxiety, depression, medical illnesses, trauma and stress related issues. Her devotedness as a therapist is inspired by personal experiences with her son. Terrence Jr. was diagnosed with TSC at three days old. Terrence's parents are both actively engaged in all aspects of Terrence's care. TSC for Terrence is accompanied with Epilepsy, Chronic Kidney Disease, Multiple Rental Cysts, Rhabdomyoma, Hypertension, Anxiety and Autism. The Hamilton family first became involved with the TSC Alliance by hosting Terrence World, A Tuberous Sclerosis Complex (TSC) Awareness and Fundraising Event. Terrence World Foundation also provided scholarship funding for students majoring in the medical profession or special needs programs. Latrice volunteered as TSC Alliance Educator Mentor with TSC Connect. The Hamilton family shared Terrence's journey with TSC at the 2018 World TSC Conference, and his story has been featured as "TSC in the News" in 2013, 2017 and 2021. He has been featured in Perspective Magazine and on TSC Navigator. Latrice was featured on the TSC Alliance's TSC Now Episode 27, sharing tips on how parents can support their children and themselves amidst the ongoing COVID 19 pandemic. Latrice was also featured in the TSC Waves Newsletter for adults with TSC in August 2021. The Hamilton family participated in the March on Capitol Hill to gain support for the TSCRP. The Hamilton Family currently hosts support events on "Hope in TSC and Autism." Latrice currently serves on the TSC Alliance's Equity, Diversity and Inclusion (ED&I) Task Force. On any given day, you will find the Hamilton Family together, laughing, dancing, exercising, boxing and acting out scenes from all the Rocky Balboa and Creed movies. Latrice and her family are passionate about living and enjoying life with hope, no matter how complex.



Karina Helboe, Chairman, Danish TSC Association, Tebstrup, Arhus, Denmark

As a spouse and a mother battling TSC since 2014, Karina lives the life of what TSC means on a daily basis and since she's a stay-at-home-mom, it's a 24/7/365 job. Each day, she navigates the world of TSC as her children are transitioning to young adulthood or already adults in a difficult world when you are different. She has worked as a volunteer in the Danish TSC association since the beginning and has chaired it for the past four years. She works hard to get Danish doctors to care about TSC since TSC Clinics are not available in Denmark. Karina also works closely with TSC Alliance and the E-TSC to support the Danish community.

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Lisa Henske, MD, Director, Center for LAM Research and Clinical Care, BWH, Harvard Medical School, **Boston MA**

Elizabeth (Lisa) Petri Henske is the Director of the Center for LAM Research and Clinical Care at Brigham and Women's Hospital. She is Professor of Medicine at Harvard Medical School, an Associate Member of the Broad Institute of MIT and Harvard and a practicing medical oncologist at the Dana-Farber Cancer Institute. She earned her undergraduate degree summa cum laude from Yale University, where she majored in Molecular Biophysics and Biochemistry, and her MD from Harvard Medical School. Dr. Henske completed a residency in Internal Medicine and a fellowship in Hematology/Oncology at the Massachusetts General Hospital. Her research laboratory is focused on the cellular, metabolic and immunologic mechanisms underlying the pathogenesis of TSC, Birt Hogg Dube and chromophobe RCC. She has most recently studied the role of FLCN and TSC in the localization and activity of TFEB (Nat Comm 2021). She is a member of the American Society for Clinical Investigation and the Association of American Physicians and serves on the Board of Directors of The LAM Foundation and the Professional Advisory Board of the TSC Alliance. Dr. Henske has received awards for her research from the TSC Alliance, The LAM Foundation, the American Thoracic Society and the Society for Women's Health Research (the Medtronic Prize).



Billy D. Holcombe, PhD, Pediatric Neuropsychologist, Le Bonheur Children's Hospital/University of Tennessee Health Science Center

Dr. Billy D. Holcombe is a pediatric neuropsychologist and director of clinical training at Le Bonheur Children's Hospital. He holds an assistant professor rank in the UTHSC Department of Pediatrics and is very committed to research and teaching across medical disciplines. Dr. Holcombe completed clinical rotations at Children's National Medical Center (Washington, DC) and Kennedy Krieger/Johns Hopkins School of Medicine (Baltimore, MD) before continuing on with residency at Children's Hospital of Michigan and fellowship at Akron Children's Hospital. Dr. Holcombe serves on several hospital committees and is an active member of the American Epilepsy Society, TSC Alliance and the National Academy of Neuropsychology.



Alex and Lesley Holmes, TSC Parents, Los Angeles, CA

Alex and Lesley are the parents of 3-year-old Seraphina, who was diagnosed with TSC before birth. Along with their son Alexander, the family lives in Los Angeles. Alex is an entrepreneur focused on media and entertainment, and Lesley is an attorney specializing in business litigation. Lesley is also Vice Chair of the TSC Alliance of Southern California and serves on the Comedy for a Cure and Step Forward to Cure TSC planning committees.





John C. Hulbert, MBBS, FRCS, FACS, Urologic Surgeon, Minneapolis, MN

John was born in England, educated at Kingswood School, Bath, UK, and attended Medical School in London, graduating MBBS in 1975. He had an internship in the UK, conducted research at University of Western Australia, in Perth, Australia and Flying Doctor in North West Australia. He received general surgical training beginning in UK and then moved to the USA for Urologic Surgery residency at University of Wisconsin and fellowship at University of Minnesota, completed in 1984; joined full time faculty at University of Minnesota, developing both clinical and basic interests and research into less invasive renal surgery for renal stones and renal cancer, with the development of a cellular injury lab, introducing low temperature treatment of renal cancer. Appointed Chair of Department 1996-2001, he then moved into private practice, continuing his academic interests. His son, Trevor, was born in 1985 and developed infantile spasms at 2 months of age, seen by Dr Gomez at Mayo Clinic and diagnosed with TSC, ultimately requiring neurosurgical treatment after which seizures were controlled. Trevor is now 36 and non-verbal and autistic, but the cornerstone of their family. John has clinical and research interests in TSC, devoting a significant part of his urologic practice to the management of the renal effects of TSC, lecturing extensively and publishing on the subject. This has led to close association with diverse groups of physicians, basic scientists, parents and caregivers with wide interests in TSC and a close association with the TSC Alliance, in particular with Dr. Michael Frost, pediatric neurologist in St. Paul, MN, co-founding and co-directing, the TSC Clinic of the Upper Midwest in the Twin Cities with him. Now recently retired, he continues in an advisory capacity with both the TSC Alliance Scientific Advisory Board and with a clear insight as Trevor's father of the issues and obstacles parents, family memers and caregivers of those with TSC face.



Emilie Hyde, TSC Alliance Adult Regional Coordinator, Owasso, OK

Emilie Hyde is a young adult with TSC. She is involved with the TSC Alliance where she is currently serving as a Adult Regional Coordinator and was a TSC Future Leader. She has been a strong advocate for others with disabilities and has attended March on Capitol Hill. She met her husband, Foster Hyde, at the 2018 World TSC Conference. Foster is also a young adult with TSC. She is a graduate of Tulsa Community College with an associate's degree in Pre-Social Work. Emilie works as a Patient Service Representative for a local medical facility.



Foster Hyde, TSC Alliance Adult Regional Coordinator, Owasso, OK

Foster Hyde is 25 years old and was diagnosed with TSC at the age of 18 months and had brain surgery at the age of 5. He was seizure free until the age of 19 when he had a breakthrough seizure. At the 2018 World TSC Conference, he met his eventual wife, Emilie, who also has TSC. They both have struggled a lot in are life but have overcome much together.



Anna Jansen, MD, PhD, Head of Pediatric Neurology, Antwerp University Hospital, University of Antwerp, Belgium

Prof. Anna Jansen is a pediatric neurologist at Antwerp University Hospital. She is affiliated to the Translational Neurosciences Research Group at the University of Antwerp and to the Neurogenetics and Mental Health and Wellbeing Research Groups at Vrije Universiteit Brussel. Her work is supported by a Senior Clinical Investigator Fellowship from the Research Foundation Flanders (FWO). Prof. Jansen runs a multidisciplinary TSC clinic and has participated in several clinical trials in TSC. Her research focuses on TSC-associated neuropsychiatric disorders. Together with Prof. Petrus de Vries, she co-leads the TANDem project, an international multi-disciplinary mobile-health project to empower families and individuals who live with TSC around the world. She serves as a scientific advisor to be-TSC, the Belgian TSC advocacy association.

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Shafali Spurling Jeste, MD, Chief, Division of Neurology, Neurological Institute Co-Director, Las Madrinas Chair, Children's Hospital, Los Angeles, CA

Dr. Jeste is a behavioral child neurologist specializing in autism and related neurodevelopmental disorders. She is Professor of Pediatrics and Neurology at the USC Keck School of Medicine and the Las Madrinas Chair, Chief of Neurology and Co-Director of the Neurological Institute at CHLA. After earning a BA in philosophy from Yale University in 1997 and her MD from Harvard Medical School in 2002, Dr. Jeste completed a residency in child neurology and a fellowship in behavioral child neurology at Boston Children's Hospital. She joined UCLA in 2010. Dr. Jeste's research is focused on developing methods to improve precision in the diagnosis and treatment of neurodevelopmental disorders. Her lab studies neurodevelopmental disorders from early infancy through late childhood. Dr. Jeste has designed innovative studies in early predictors of autism in TSC that integrate biomarkers with behavior to define atypical development prior to the onset of autism. This work in TSC has led to the first randomized controlled clinical trial of behavioral intervention for these infants and has paved the way for other early intervention trials in rare genetic syndromes. Dr. Jeste's research is directly inspired by her clinical work. To address the many gaps in medical care for rare genetic forms of neurodevelopmental disorders, she founded the CARING (Care and Research in Neurogenetics) Clinic. This clinic became the hub for several new clinical trials for genetic syndromes. Dr. Jeste's work is funded by the National Institutes of Health, the Department of Defense and the Simons Foundation. She holds several national and international leadership positions including the Board of Directors of the American Brain Foundation, Board of Directors of the National Organization for Rare Disorders and the Board of Directors of the International Society for Autism Research; she recently served as the Chair of the International Baby Siblings Research Consortium. In 2019 she was awarded the Presidential Early Career Award for Scientists and Engineers for her innovations in research in early predictors and intervention for genetic neurodevelopmental disorders.



Celie Johnson, Former TSC Alliance Future Leader, Boston, MA

Celie was diagnosed with TSC when she was 6 months old after having infantile spasms and since then has defied many of the odds given at the time. She graduated from high school, was accepted to her dream college in Boston, MA, and graduated from Boston University on the Dean's List. Celie stays pretty busy at Brigham and Women's Hospital, where she works as an MA/PA in outpatient dermatology, and is taking prerequisites for nursing school, which she will be applying to later this summer!



David and Rachel Johnson, TSC Parents, Huntsville, AL

David and Rachel Johnson were thrust into the TSC way of life three years ago before their son, Noah, was born. Rhabdomyomas were detected on a 34-week ultrasound, which quickly led them to a TSC diagnosis. Prevention has always been a huge part of Noah's story, including enrollment in the PREVeNT trial. David and Rachel started their TSC journey with a two-fold goal that has continued to guide how they have taken on TSC. Those goals are to seek the best care possible for Noah and do what they can to help positively move the goalpost for the global TSC community.



Prof. Sergiusz Jóźwiak, MD, PhD, Medical University of Warsaw, Poland

Sergiusz Jóźwiak is a professor and the head of the Department of Pediatric Neurology at Warsaw Medical University. He received his medical degree from the Medical University Warsaw in 1983 and doctoral degree from the Children's Memorial Health Institute in 1990. Prof. Jóźwiak completed his habilitation at the same institute in 1995 and was appointed Head of the Paediatric Neurology and Epileptology Department in 1997 in the same institution. He held this position until May 2015, when he moved to Warsaw Medical University. From 2009-2014 he served as a National Consultant in Paediatric Neurology. Prof. Jóźwiak's research focuses mainly on neurocutaneous disorders and epilepsy, especially infantile spasms. For more than 25 years, he has led a special program for TSC patients and worked out practical guidelines for TSC management. In 2009 Prof. Jóźwiak received the prestigious Manuel Gomez Award established by TSC Alliance for "creative or pioneering efforts that have appreciably improved either the understanding of the disease or the clinical care available for individuals with TSC." In years 2013-2019 he was a coordinator of the large-scale European Commission Project EPISTOP evaluating clinical and molecular biomarkers of epileptogenesis in a genetic model of epilepsy -tuberous sclerosis complex (www.EPISTOP.eu). Prof. Jóźwiak is an active member of numerous international organizations and has published more than 300 papers in national and international peer-reviewed journals. Prof. Jóźwiak is on the editorial boards of several professional journals, including Pediatric Neurology (USA), European J.Paediatric Neurology (Amsterdam) and Journal of Child Neurology (USA).



Carly Kaye, Community Programs Manager, TSC Alliance, Plymouth, MI

Carly joined the TSC Alliance as Community Programs Manager in April 2022 after serving as a volunteer for the past six years including Chair, TSC Alliance of Michigan, and Clinic Ambassador at the TSC Clinic at Beaumont Royal Oak. In addition, she served on several planning committees, including the TSC Alliance's 45th Anniversary Gala. Carly also began designing and making apparel and other merchandise to help members of the TSC community promote their advocacy and to raise awareness of TSC, which eventually led to creating the TSC Swag Store in 2018. Carly is a consistent, supportive and resourceful voice for families and individuals, particularly in online settings. She takes pride in and enjoys connecting people, helping those who are in need and most of all giving newly diagnosed families hope. Carly graduated from the University of Florida in 2006 with a degree in History and Business. After college she worked as a before/after school program coordinator for several years before becoming an elementary school teacher. In 2016, Carly stepped away from teaching to care for her son Logan, who was diagnosed with TSC shortly after his first birthday.



Mark R. Keezer, MDCM, PhD, Neurologist, Centre Hospitalier de l'Université de Montréal (CHUM), Associate (Clinical) Professor Department of Neurosciences & School of Public Health of the Université de Montréal, Canada

Mark Keezer is a clinician researcher at the Centre Hospitalier de l'Université de Montréal (CHUM), Associate Professor (clinical) in the Department of Neurosciences and Adjunct Professor at the School of Public Health of the Université de Montréal. Dr Keezer's clinical and research training began at McGill University, was continued at the National Hospital for Neurology and Neurosurgery, Queen Square, UK and then ended with his PhD in Translational Neuroscience at Leiden University, the Netherlands. Dr Keezer is a member of the editorial board for Epilepsia, the epilepsy surgery commission of the International League Against Epilepsy and the guidelines subcommittee of the American Academy of Neurology. Dr. Keezer is currently supervising a number of research projects focused on physical frailty and the somatic comorbidities of epilepsy. Since 2016, Dr. Keezer is the director of the CHUM TSC clinic, where he focuses on the care of adults living with TSC.

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(John) Chris Kingswood, MBBS, MSc, FRCP, Brighton & Sussex University Hospitals Trust, Renal Unit, Brighton, UK

Dr. Kingswood is a UK renal physician working St. George's Hospital medical school and the Sussex Kidney Unit, Brighton & Sussex University Hospitals Trust. He was co-founder of the multidisciplinary Tuberous Sclerosis Clinic at St George's Hospital. Dr Kingswood has published numerous papers on the natural history and treatment of TSC. He was chief UK investigator for the Exist-2 and TOSCA studies. Since 2014 he has worked 50% of the time in clinical medicine and 50% in research, concentrating on renal genetic disorders.



Mary Kay Koenig, MD, Co-Director UT TSC Center of Excellence, University of Texas McGovern Medical School, Houston, TX

Dr. Mary Kay Koenig serves as the Director of the Center for the Treatment of Pediatric Neurodegenerative Disease (CTPND) and Director of Research for the Division of Child & Adolescent Neurology at the University of Texas McGovern Medical School. Dr. Koenig attended the University of Texas in Austin where she received a Bachelor of Arts in BioChemistry. Following undergraduate school, Dr. Koenig received a Master's of Science degree in Microbiology at Southwest Texas State University followed by her Medical Doctorate at St. George's School of Medicine. After medical school, Dr. Koenig completed a pediatrics residency at the University of Texas Medical Branch in Galveston and a fellowship in Child and Adolescent Neurology at The University of Texas Medical School. Upon completion of her fellowship, she joined the faculty and is now a fully tenured professor. She has served as the director for the UT Mitochondrial Center of Excellence since 2007, the co-Director for the UT Memorial Hermann Tuberous Sclerosis Clinic since 2007 and the Leigh Syndrome clinic director since 2013. Dr. Koenig strives to provide excellent clinic care and advance research for children afflicted with genetic neurodegenerative disease. She also works to mentor and educate junior faculty, fellows, residents and medical students in the art of medicine and clinical research.



Jessica Krefting, RN, BSN, Nurse/Research Coordinator, UAB TSC Clinic, Birmingham, AL

Jessica received her Bachelor of Science from the University of Alabama Birmingham in 2004. She has been a registered nurse in pediatrics for more than 17 years with the last 14 years in the fields of TSC and epilepsy. Since 2012, she has been involved as a Research Nurse Coordinator for multiple TSC studies, has been the lead coordinator for the PREVeNT study for the last 5 years and has worked on other studies including TACERN, RDCRN, TSC-EBS, and STEPS. Jessica is also the Nurse Coordinator for the UAB TSC Clinic and coordinates the care for both the adult and children across specialists. She has been a TSC Alliance Board of Directors member since 2021 and currently serves as Secretary.



Darcy A. Krueger, MD, PhD, Clack Endowed Professor and Director, Tuberous Sclerosis Clinic, Cincinnati Children's Hospital, University of Cincinnati College of Medicine, Cincinnati, OH

Darcy A. Krueger received MD and PhD degrees from Saint Louis University (Missouri, USA) and completed pediatrics and neurology training at Cincinnati Children's Hospital (Ohio, USA). He currently is the Clack Endowed Chair in Tuberous Sclerosis and Director of the Tuberous Sclerosis Clinic at Cincinnati Children's Hospital Medical Center. He is also Professor of Clinical Pediatrics and Neurology at the University of Cincinnati College of Medicine. The TSC Clinic at Cincinnati Children's Hospital provides comprehensive, multidisciplinary clinical care to TSC patients of all ages and is recognized as a leader in clinical and translational research aimed at developing new treatments for individuals affected by TSC. Dr. Krueger is currently the lead investigator for TSC-STEPS, the first prospective multicenter clinical trial evaluating sirolimus treatment during infancy to prevent future progression of neurological manifestations of TSC, including epilepsy and neurodevelopmental disabilities. For contributions in TSC research and patient care, Dr. Krueger was recipient of the Manuel R. Gomez Award in 2019. Dr. Krueger is a member of the TSC Alliance Board of Directors, actively serving on committees focused on global outreach, advancing research, and improving delivery of TSC patient clinical care.



Mary Ann Pregnall Lamb, PhD, TSC Alliance Volunteer, Durham, NC

Mary Ann is the proud grandmother of four amazing grandchildren, one of whom, Jay Lamb, has TSC. Prior to her retirement, Mary Ann spent her career researching rare blood coagulation and plasma protein deficiency disorders and ensuring the licensure of orphan drug products to treat these rare disorders. Her career spanned across academia (UNC Chapel Hill), the federal government (FDA) and the biopharmaceutical industry (Bayer/Talecris/Grifols/BPL). Since retiring, Mary Ann has made it her passion to advocate for continued federal funding of the Tuberous Sclerosis Complex Research Program (TSCRP) and to fundraise to support the TSC Alliance's research and awareness programs, especially programs that enhance quality of life for TSC warriors. Mary Ann is excited about the opportunity to engage with other TSC grandparents to find ways to further support families and communities.



Julian A. Martinez-Agosto, MD, PhD, Associate Professor, UCLA, Los Angeles, CA

Dr. Julian A. Martinez-Agosto is a medical geneticist and developmental biologist who has served on the UCLA School of Medicine faculty since he joined the Department of Human Genetics in 2007. Dr. Martinez earned his BS in Biology and Spanish Literature at Yale University and pursued his medical training at Yale School of Medicine. He went on to train as a pediatrician at Mattel Children's Hospital UCLA. Dr. Martinez-Agosto was Chief Resident of the UCLA Intercampus Medical Genetics Program, where he trained as and currently is a board-certified Medical Geneticist. Dr. Martinez is a recipient of the Pediatric Department Outstanding Research Award, as well as the David W. Smith Pediatric Trainee Research Award of the Western Society for Pediatric Research, among other honors. Dr. Martinez-Agosto is a member of the Division of Medical Genetics, where he has maintained a clinical interest in genetic syndromes that lead to neurodevelopmental differences, overgrowth, vascular malformations, and cancer predisposition. His laboratory research focuses on novel growth regulatory pathways in progenitor and stem cell maintenance and the study of human growth disorders associated with neurodevelopmental phenotypes and cancer predisposition.



Kirk McCreary, Former TSC Alliance Future Leader, Denver, CO

Kirk McCreary is a young adult who was diagnosed with TSC at two years of age. Kirk graduated from the University of Redlands with bachelor's degrees in Global Business and German in April 2020. After graduation, Kirk worked in the supply chain and logistics industry until he transitioned into a career of teaching business at the high school level. During the 2020 to 2021 calendar year, Kirk served as a Future Leader with the TSC Alliance where he, alongside his fellow Future Leaders, produced and presented a series of webinars focusing on young adult TSC-related topics. Kirk is both eager and excited to continue working with the TSC Alliance and looks forward to connecting those both directly and indirectly affected by TSC, as well as continuing to foster greater awareness of TSC.



Melyssa McDonough, Director, Behavior Supports for Northway Academy–Community Behavior Supports, Brooklyn Park, MN

Melyssa McDonough is the Director of Behavior Supports for Northway Academy–Community Behavior Supports. She provides intensive community-based behavior analysis for adults and adolescents living in a residential community-based setting. Melyssa supervises more than 10 behavior analysts in their service delivery as well as maintains a caseload of her own. In her spare time Melyssa is a long-distance runner, wife and mom to two very active young children. Melyssa was diagnosed with TSC and LAM when she was 29 years old and is able to offer a unique perspective of a clinician and someone living with the effects of the disease and serves as an Adult Regional Coordinator with the TSC Alliance..

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Shelly Meitzler, Director, Community Education & Resources, TSC Alliance, Silver Spring, MD

Shelly joined the TSC Alliance in January 2017 after spending 14 years volunteering in the TSC community, including serving as Community Alliance Chair, Step Forward to Cure TSC Chair in Delaware/Lehigh Valley and Education Parent Mentor. Her background is business management. Her drive and passion have been fueled by her two children with TSC, Ashlin and Mason, and her daughter, Mikenna. Experiencing the need to advocate educationally and medically for her family when their journey began with TSC two decades ago, Shelly realized she wanted to work alongside and empower TSC families by providing resources and support to advocate for themselves and their children to secure the services they are entitled to receive and to realize their full potential. Shelly completed the Special Education Advocate Training 1.0 and 2.0 through the Council of Parent Attorneys and Advocates. She supports TSC families, conducts school meetings and TSC training sessions and provides tremendous insight as a parent having walked the TSC journey - twice.



Joanna Lomas Mevers, PhD, BCBA-D, Director, Severe Behavior, School Consultation and Care Coordination Programs, Marcus Autism Center/Associate Professor, Emory School of Medicine, Department of Pediatrics, Atlanta, GA

Dr. Lomas Mevers is the current Director for the Severe Behavior, Care Coordination and School Consultation Programs at the Marcus Autism Center in Atlanta, GA. She is also an Associate Professor in the Department of Pediatrics at Emory University School of Medicine. Dr. Lomas Mevers completes her clinical work in the Severe Behavior Program where she specializes in the assessment and treatment of severe challenging behaviors and toileting training for children diagnosed with developmental disabilities such as autism spectrum disorder. In addition to her clinical work, she also conducts research in these areas. She has published 30 research articles focused on the assessment and treatment of challenging behavior and toilet training. The Marcus Autism Center's toileting program sees more than 50 kids per year across the three different toileting programs which includes two programs that focus on enuresis and one program that focuses on encopresis. She has more than 20 years of experience with toilet training individuals with autism spectrum disorders (ASD) and other related disabilities. In addition, she has been awarded over \$1 million in grants to further the advancement of a multidisciplinary treatment for encopresis for children with ASD. She has also published on this topic and continues to focus much of her research on the advancement of toilet training for children with developmental disabilities.



Debora Moritz, TSC Alliance Volunteer, Scottsdale, AZ

Debora Moritz is mom to Griffin, a young man with TSC. She is a volunteer who focuses on advocacy, scientific literacy and improving the daily lives of people living with TSC. Debora has volunteered with the TSC Alliance as a Board member and serving as the Chair of the Government Relations Committee focusing on developing grass roots advocates supporting continued funding of the TSCRP. In Arizona she is available help constituents find resources and connect with other families living with TSC.



Robert Moss, President, Seizure Tracker LLC, Springfield, VA

Robert Moss knows first-hand how it feels to have a child with TSC and epilepsy. It was the helplessness he felt in his own situation that inspired him to create Seizure Tracker. He watched his son, Evan, have countless seizures and knew there had to be a more effective way to monitor the seizure activity and medications than writing them down on paper. Putting his commercial photography business aside and teaching himself to be a website developer, he began the career shift that would ultimately provide SeizureTracker.com to a global community. Seizure Tracker is a free resource with more than 35,000 users in as faraway places as Sweden, Japan, Qatar and India. Robert is dedicated to empowering people living with epilepsy and giving back to the community he is also very much a part of.



Joel Moss, MD, PhD, Senior Investigator and Deputy Chief, Pulmonary Branch, NHLBI, NIH, Bethesda, MD

Joel Moss, MD, PhD, is Deputy Chief and Senior Investigator in the Pulmonary Branch, NHLBI, NIH and Head of the Translational Research Section and the Lymphangioleiomyomatosis (LAM) program. He graduated Brandeis University, summa cum laude, and received MD and PhD degrees from New York University School of Medicine. He is a member of Phi Beta Kappa and Alpha Omega Alpha. Following his internship and residency (Medicine; Johns Hopkins), he completed post-doctoral and pulmonary fellowships (NHLBI). He has co-authored more than 800 scientific papers, edited/co-authored books and is co-inventor of patents. Dr. Moss was a member of the NHLBI Institutional Review Board from 1988-2006, Chair from 1995-2006 and co-authored texts on ethical considerations in clinical protocols. Dr. Moss has received multiple awards, including The LAM Foundation awards, the John W. Walsh PAR Award for Excellence from ATS and the New York University Alumni Achievement Award in Basic Science and the Chiba (Japan) Medical Society Award. Dr. Moss is an honorary member of the Peruvian Thoracic Society. He is an elected member of the Association of American Physicians and the American Society for Clinical Investigation (ASCI) and served as ASCI Councilor and Vice-President. Dr. Moss was elected to the Johns Hopkins University Society of Scholars (2009). He is a Fellow of the American College of Chest Physicians and the American Thoracic Society. Dr. Moss was an editor of the Journal of Biological Chemistry, Journal of Clinical Investigation and the European Respiratory Review. He is currently an editor of CHEST and the American Journal of Respiratory Cell and Molecular Biology. His clinical program focuses on LAM and cystic lung diseases. LAM results from proliferation of LAM cells and occurs with increased frequency among women with TSC. Dr. Moss and colleagues initially established the natural history of LAM with detailed clinical phenotyping of LAM and TSC patients and characterized the phenotypic and genotypic features of LAM lung.



Hope Northrup, MD, Director and Professor, Division of Medical Genetics, Department of Pediatrics, McGovern Medical School, UTHealth, Houston, TX

Hope Northrup, MD, is a Medical Geneticist (Clinical, Biochemical and Molecular) in the Department of Pediatrics at the McGovern Medical School, UTHealth, Houston. She is Professor and Director of the Division of Medical Genetics. Dr. Northrup is the Director of the Medical Genetics Residency/Fellowship Training Program (1993-present). She has been selected numerous times for inclusion in various publications citing excellence in clinical care including Best Doctors in America, America's Top Doctors and Texas Super Doctors. In recognition of her teaching excellence, she received the Regents Outstanding Teaching Award for the University of Texas System in 2016. Dr. Northrup received the Excellence in Clinical Science Award for 2020 from the McGovern Medical School Women Faculty Forum, Dr. Northrup obtained an MD degree from the Medical University of South Carolina. She completed a Pediatric Residency at Children's Medical Center/Southwestern Medical School in Dallas, Texas, and a Medical Genetics Fellowship at the Institute for Molecular Genetics, Baylor College of Medicine in Houston, Texas. Dr. Northrup's research interests focus on unraveling the basis of neurogenetic diseases, specifically TSC and spina bifida and developing therapies for genetic diseases. She was the 2003 winner of the Manuel R. Gomez Award from the TSC Alliance for her research and clinical contributions in the field of TSC. Dr. Northrup has multiple research projects funded through NIH, private foundations and industry. She has authored/co-authored more than 160 peer-reviewed articles. Dr. Northrup has provided numerous national and international invited presentations.

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Angela Peron, MD, PhD, ASST Santi Paolo e Carlo, San Paolo University Hospital, Milan, Italy

Angela Peron is a clinical geneticist. She earned her MD degree from Vita-Salute San Raffaele University in Milan in 2009, completed a residency in Medical Genetics at the University of Milan in 2015 and earned her PhD in Clinical Research at the University of Milan in 2021. She is currently an attending physician in medical genetics at San Paolo Hospital in Milan (Italy) and an adjunct assistant professor at the University of Utah (USA). She serves as the geneticist of the pediatric and adult TSC clinic of San Paolo University Hospital in Milan and also provides clinics for children and adults with different genetic conditions. Her primary research interests focus on individuals with TSC and No Mutation Identified, mosaicism in TSC, and genotype-phenotype correlations. She is also involved in multiple research studies on epilepsy and autism in TSC. In 2017 she organized the 2nd Early-Stage Tuberous Sclerosis Complex Researcher Meeting together with four other young researchers and was appointed co-chair of the Early Career Researcher Symposium held in 2019. She was part of the scientific committee of the 2020 International Tuberous Sclerosis Complex Research Conference. Her other research interests include dysmorphology, genetics of epilepsy and autism spectrum disorder and genomic medicine.



M. Scott Perry, MD, Head of Neurosciences, Cook Children's Medical Center, Fort Worth, TX

Dr. M. Scott Perry is the Head of Neurosciences and the Director of the Genetic Epilepsy Clinic at the Jane and John Justin Neurosciences Center at Cook Children's Medical Center, Fort Worth, TX. He completed his MD at the University of Mississippi School of Medicine and general pediatrics and child neurology training at Emory University. He completed a Clinical Neurophysiology fellowship at Miami Children's Hospital before joining Cook Children's in 2009. His clinical and research interests include the use of epilepsy surgery for the treatment of intractable childhood epilepsy and the evaluation and treatment of genetic epilepsy syndromes.



Steven Peters, TSC Parent, TSC Alliance Volunteer, Owasso, OK

Steven Peters is a volunteer for the TSC Alliance where he has served as a Community Alliance Chair, participated in the March on Capitol Hill and supported many TSC Alliance fundraising efforts. Steven and his wife, Kristy, live in Owasso, OK and have two married daughters. Their youngest daughter, Emilie, age 25 is affected with TSC. Among the many challenges of TSC, Steven has spent time navigating the transition for his daughter from adolescence in public schools, through college and into the workforce. Steven owns and operates an insurance agency. He enjoys spending time with his family, playing golf, and collecting cast iron cookware.



Emily Phan, TSC Alliance Future Leader, Malibu, CA

Emily Phan is a world-class pianist living with TSC. At 6 months old, Emily suffered from infantile spasms and was subsequently diagnosed with TSC, which caused major cognitive and fine motor skill developmental delays. However, this never stopped her from fighting to live a normal life. With the guidance and support of the TSC Alliance, her mother was able to find the best medical care for Emily. To assist with normal development, Emily had five different piano teachers, which ultimately supplemented her speech, occupational and physical therapy. At the age of 5, she had a dream of spreading beauty and love by utilizing the sound of the piano as her medium. Over the years, Emily caught up with her age milestones. She began competing in district, regional and national-level piano competitions. Moving on to college, she was accepted to Pepperdine University on a talent-based scholarship, graduating with her Peformance degree in 2019. Emily is the recipient of numerous first place piano awards, which have led to invitations to perform across Europe, Russia and the United States. Emily has performed in renowned concert halls such as Carnegie Hall in NYC, Mozarteum Concert Hall in Austria and the Richard Nixon Library in California. Today, Emily inspires families, TSC patients and other musicians globally. She aspires to give hope and share her light with other families afflicted by TSC. Emily closed Comedy for a Cure 19.5 with a riveting performance that brought everyone to their feet.



Kelly Piacenti, MA, ChSNC®, Head, MassMutual SpecialCare, Springfield, MA

Kelly Piacenti is responsible for the MassMutual SpecialCare program, which includes nearly 500 Special Care Planners. She also oversees partnerships between MassMutual and the largest special needs non-profit organizations across the country, including The Arc, Autism Society, Huntington's Disease Society of America, National Down Syndrome Congress, TSC Alliance and United Cerebral Palsy (UCP). Kelly serves on the National Board of Directors for UCP and was formerly on the National Board of The Arc for 8 years until 2020. She also serves on the Advisory Board for The Academy of Special Needs Planners as well as The American College MassMutual Center for Special Needs. Kelly has spoken at dozens of industry conferences across the country and has been featured as a speaker on several radio talk show programs. Kelly is also published and quoted in numerous industry publications for the special needs community. In May 2017, the Theresa Foundation awarded Kelly as the recipient of the annual 2017 Theresa Award for her outstanding community service and advocacy on behalf of people with special needs. And in the fall of 2015, Kelly was awarded the distinction by "Working Mother" magazine, as a "Working Mother of the Year" in their "100 Best Companies" featured article. She has worked with MassMutual since July of 2016. Before joining MassMutual, she led the MetLife Center for Special Needs Planning, where she was employed from 1991 through 2016. Kelly holds a master's degree and resides in Chester, NJ with her husband and children. She was the mother of a child with special needs for 19 years and possesses the knowledge, insight and empathy to act as a mentor and resource for families facing similar challenges.



Brenda Elaine Porter, MD, PhD, Professor of Neurology, Stanford University, Stanford, CA

Dr. Porter obtained her undergraduate, MD and PHD degree at Washington University in St. Louis. She currently is Professor of Neurology at Stanford, Director of Pediatric Epilepsy at Lucile Packard Children's Hospital and co-director of the Tuberous Sclerosis Complex Clinic at Stanford. Her research is focused on the role of the EEG as a biomarker in tuberous sclerosis complex, developing therapies for the prevention of epilepsy and improving neuronal development in TSC. Her passion is trying to understand the role and timing of epilepsy surgery for providing the best long-term outcomes for children with TSC. She enjoys mentoring residents, fellows and junior faculty at Stanford and through the NIH Child Neurologist Career Development K-12 Award Program (CNCDP-K12).



Ashley Pounders, MSN, FNP-C, Director, Medical Affairs, TSC Alliance, Memphis, TN

Ashley joined the TSC Alliance in 2020. She recently led the development of the TSC Alliance's new online "TSC Navigator" tool to help individuals and families maneuver the intricate TSC healthcare system and proactively manage their care. She obtained her MS in Nursing in 2018 from Purdue University and her BS in Nursing in 2007 from Baptist College of Health Sciences in Memphis. Ashley served as the lead coordinator for the TSC Center of Excellence at the LeBonheur Children's Hospital in Memphis from 2016 through 2019. In 2018, Ashley played a major role in assisting with the inaugural TSC Nursing Conference in Dallas. Ashley also was part of many clinical trials and retrospective research projects. She received a Daisy Award during her time at LeBonheur for facilitating process and compassionate changes to better assist the TSC patient population.



Emily Racenet, TSC Alliance Future Leader Mentor, Wasilla, AK

Emily has been around TSC since her younger sister was born. Her family didn't know of her condition until well after she was born when they started to notice her staring off at ceiling fans and other odd objects and realized these were absence seizures. Flash forward 16 years to today, Emily spends a large portion of her time working with the TSC Alliance or on other TSC-related matters, from the Future Leaders program to interviews with Amy Poehler's Smart Girls about the condition to proposing resolutions to continue funding for the TSCRP to the largest youth leadership conference in Alaska, the Alaska Association of Student Governments.

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Raj Rajaraman, MD, MS, Assistant Professor, UCLA, Los Angeles, CA

Rajsekar "Raj" Rajaraman, MD, MS, is an Assistant Professor in Pediatric Neurology at the UCLA Mattel Children's Hospital. He completed his Pediatric Neurology residency, Clinical Neurophysiology fellowship and Epilepsy fellowship training at UCLA. Currently, his clinical and research interests involve all aspects of infantile spasms, as well as its effects in TSC and CDKL5 Deficiency Disorder. Dr. Rajaraman is Director of the UCLA Tuberous Sclerosis Center of Excellence as well as the UCLA CDKL5 Center of Excellence. He is the primary investigator of more than a dozen pharmaceutical and investigator-driven clinical trials and studies. He has presented research at multiple conferences, including the American Academy of Neurology, Child Neurology Society and the American Epilepsy Society, receiving achievements including the national 2014 American Epilepsy Society Young Investigator Award. Dr. Rajaraman has multiple publications on infantile spasms and cannabidiol (CBD). In addition to his clinical and research interests, Dr. Rajaraman is an active member of the American Academy of Neurology (AAN) and American Epilepsy Society (AES). He volunteers for the Epilepsy Foundation of Greater Los Angeles, including helping at their annual Walk to End Epilepsy at the Rose Bowl, as well as the Epilepsy Awareness Day at Disneyland.



E. Steve Roach, MD, Professor of Neurology, University of Texas Dell Medical School, Austin, TX

E. Steve Roach, MD, is Professor of Neurology at the University of Texas Dell Medical School in Austin, TX, where he serves as the Associate Chair for Operations of the Department of Neurology and as Chief of the newly created UT Health Austin Pediatric Neuroscience Center at Dell Children's Hospital. Dr. Roach has written or edited 10 books and has published numerous articles, commentaries and chapters, including many articles focusing on the clinical manifestations of TSC and other genetic disorders. Dr. Roach is a past president of the Child Neurology Society and recently concluded a nine-year term as the editor-in-chief of Pediatric Neurology.



Steven L. Roberds, PhD, Chief Scientific Officer, TSC Alliance, Silver Spring, MD

Steven Roberds joined the TSC Alliance staff in 2011 as Chief Scientific Officer. In this role, Steve leads the development and execution of the TSC Alliance's research strategy through partnerships and conversations with all stakeholders, including individuals and families affected by TSC, basic and clinical researchers in academia and industry, healthcare providers, government agencies involved in medical research, and other non-profit organizations. Steve led the implementation of the 2012 TSC Clinical Consensus Conference and an update to the consensus guidelines in 2021, Together with the CEO, CFO and Board, he developed a seven-year Research Business Plan beginning in 2019 to grow the TSC Alliance's research programs, including a Preclinical Consortium to accelerate testing of potential new treatments, a Biosample Repository to collect and share biosamples from individuals enrolled in the TSC Natural History Database, a Clinical Research Consortium, a TSC Learning Healthcare System, Innovation Workshops, conferences, and grants. Steve was awarded his PhD in Pharmacology from Vanderbilt University in 1992, after which he was introduced to rare disease research as a postdoctoral fellow at the University of lowa studying the biology and genetics of muscular dystrophies. He then began a 16-year research career in the pharmaceutical industry. Immediately prior to joining the TSC Alliance, Steve was an Associate Research Fellow and project leader responsible for driving global project teams toward new human proof-of-concept studies to repurpose Pfizer compounds for new indications.



David F. Rodriguez-Buritica, MD, Associate Professor, Division of Genetics, Department of Pediatrics, McGovern Medical School, The University of Texas Health Science Center at Houston, TX

Dr. Rodriguez-Buritica is a board-certified pediatric endocrinology and medical geneticist. He attended medical school at his home country Colombia, South America and then moved to the US and pursued training in General Pediatrics and Pediatric Endocrinology at Winthrop University Hospital in New York, followed by a Medical Genetics Fellowship at the Department of Genetics in University of Alabama at Birmingham. He joined the Division of Medical Genetics at the Department of Pediatrics, McGovern Medical School of the University of Texas at Houston Medical Center in 2014. He specialized in carbohydrate inborn errors of metabolism, hypoglycemia, Disorders of Sexual development, genetic forms of Rickets, endocrine abnormalities in TSC, among other topics that combined genetics and endocrinology. He directs the Glycogen Storage Disorder center of excellent at the McGovern Medical School and performs endocrine evaluation for TSC patients at UT TSC Center of Excellence.



Kari Luther Rosbeck, President and CEO, TSC Alliance, Silver Spring, MD

Kari Luther Rosbeck joined the TSC Alliance in June 2001 and became President and CEO in November 2007. She has been involved in nonprofit fundraising and volunteer management for more than 30 years. During Kari's tenure as President and CEO, the TSC Alliance established a comprehensive research program fostering collaboration with industry and academia to move treatments for TSC forward in a more expedited way. The "Unlock the Cure" research strategy focuses on key points along the drug discovery path and has also served as a capital fundraising campaign with more than \$19 million raised for TSC research since August 2011. Because of her leadership, the organization has taken an active role in educating the TSC community about clinical trials to diminish the time for recruitment, including pivotal trials that have led to three FDA-approved drugs specifically for TSC. Kari previously served as Executive Vice President, overseeing the national volunteer outreach program and was responsible for fund development. She developed and implemented a vast national network of more than 30 volunteer branches called Community Alliances; increased volunteer participation from 90 individuals to more than 2,000; and through her involvement in special events and major gift fundraising, helped increase the annual revenue by more than 50 percent.



Micaela Rozenberg, President, Tuberous Sclerosis Association in Portugal (AETN), Portugal

Micaela Rozenberg has two daughters; the youngest who is now 26 has TSC. Micaela has been the President of AETN, the Portuguese TSC association, which she founded when her daughter was 16 years old. She understood how much parents and patients miss when they have no involvement in the association's initiatives. Micaela is also Co-Chair of The European TSC association (ETSC) and on the board of directors of RD-Portugal (the Portuguese Rare Diseases Federation). Both AETN and ETSC just celebrated their 10th anniversaries. Micaela is also involved in the TSC international community and also advocates passionately to improve care for TSC patients and their families from around the world by empowering both the patient community and the health care professionals. She has been involved in creating national, European and worldwide networks between key opinion leaders and local doctors. She has been leading the ETSC DEA project (Disseminating Expertise and Achievement) connecting Europe both on medical and patient level. She advocates fiercely for inclusion to ensure no family is left out.

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Gabrielle Rushing, PhD, Associate Director, Research, TSC Alliance, Nashville, TN

Gabrielle joined the TSC Alliance staff in December 2019 and works remotely from Nashville, TN. Her efforts focus on stimulating innovative research and leading the Natural History Database and Biosample Repository projects. She completed her PhD at Vanderbilt University, studying TSC brain tumor development and neural stem cells under the guidance of Dr. Rebecca Ihrie. Prior to joining the TSC Alliance, Gabrielle served as Project Manager for the PREDICT (Pharmacogenomic Resource for Enhanced Decisions in Care and Treatment) program at Vanderbilt University Medical Center. Before joining the staff, she completed an internship at the TSC Alliance in 2017, where she participated in the March on Capitol Hill, contributed to the Patient-Focused Drug Development meeting and wrote the research presentation summaries at the International TSC Research Conference. Additionally, Gabrielle served as the Research Director for the Thisbe and Noah Scott Foundation from 2016-2019.



Carlie Russell, TSC Alliance Volunteer, Coppell, TX

Carlie Russell is a 26-year-old graduate of Texas A&M University with a degree in Communication. Her younger sister Anna was diagnosed with TSC at birth. Carlie currently works for Spire Hospitality as its Corporate Human Resources Director. She has been involved with the TSC Alliance since she graduated college and moved back to the Dallas/Fort-Worth area.



Leslie Russell, TSC Alliance Volunteer, Flower Mound, TX

Leslie Russell lives in Flower Mound, Texas and is a mother of two girls—Carlie, 26, and Anna, 24, an adult living with TSC. Anna was diagnosed with TSC at 8 months old. Leslie has been involved with the TSC Alliance for more than two decades and currently serves as the Chair of the TSC Alliance of the Dallas/Ft. Worth Area and a Dependent Adult Transition Coordinator. She also served on the Treasure Street Board at Texas Scottish Rite Hospital for Children. She and her husband Chris have supported research to improve the quality of life for Anna and all other individuals with TSC. They are both also currently volunteers and supporters of 29 Acres, which provides a safe and dynamic supported living community where adults with ASD and other neuro-diversities live, enabling them to build relationships and to feel value, success and happiness in their lives.



Mustafa Sahin, MD, PhD, Director, RSZ Translational Neuroscience Program, Boston Children's Hospital, Harvard Medical School, Boston, MA

Mustafa Sahin, MD, PhD, is Professor of Neurology at Harvard Medical School and Director the Rosamund Stone Zander Translational Neuroscience Center at BCH. He has established and directs the Multidisciplinary Tuberous Sclerosis Program at Boston Children's Hospital. The research in the Sahin laboratory is directed at understanding the cellular mechanisms of neuronal connectivity and their relationship to neurological dysfunction. His research focuses on TSC and related neurodevelopmental disorders using cell and animal models as well as clinical investigations. He is the principal investigator of the Rare Diseases Clinical Research Network (RDCRN), studying the comparative pathobiology of TSC, PTEN and SHANK3 mutations in patients as well as co-PI of the BCH/HMS Intellectual and Developmental Disorders Research Center (IDDRC). He serves on the TSC Alliance Board of Directors and chairs both the Science and Medical Committee and the International Scientific Advisory Board.



Joshua Samuels, MD, MPH, Professor of Pediatrics and Medicine, Division of Pediatric Nephrology and Hypertension, McGovern Medical School at the University of Texas in Houston, TX

Joshua Samuels, MD, MPH, is a tenured Professor of Pediatrics and Medicine in the Division of Pediatric Nephrology and Hypertension at the McGovern Medical School at the University of Texas in Houston. A native Houstonian, Dr. Samuels received his medical degree from Baylor College of Medicine and a Combined Internal Medicine/Pediatric Residency at the University of Rochester. He completed a combined adult and pediatric nephrology fellowship in 2003 at the UT Health Science Center at Houston and has remained on faculty ever since. Dr. Samuels' clinical and research interests are in the fields of pediatric hypertension and TSC. He is the clinical nephrologist for the University of Texas Memorial Hermann Tuberous Sclerosis Center of Excellence in Houston, where he evaluates and treats both children and adults with renal complications of TSC. Dr. Samuels serves on the Professional Advisory Board of the TSC Alliance. Dr. Samuels has published more than 80 peer-reviewed scholarly articles with more on the way. He is a Fellow of the American Academy of Pediatrics, American Society of Nephrology, and the American Heart Association. As a teacher, Dr. Samuels is fellowship director of the UT Houston Pediatric Nephrology and Hypertension training program and a member of the school's curriculum committee. He gives lectures for medical students, residents, fellows and faculty not only covering nephrology topics but also epidemiology, critical appraisal and evidence-based medicine. He is a member of the McGovern Medical School's Academy of Master Educators, has received multiple Dean's teaching awards and has been an invited lecturer about both pediatric hypertension and TSC at both national and international meetings.



Mindy Scheithauer, PhD, BCBA-D, ABPP, Associate Professor, Emory University School of Medicine, Psychologist, Marcus Autism Center, Atlanta, GA

Dr. Scheithauer is a licensed psychologist and doctoral-level board-certified behavior analyst who is board certified in behavioral and cognitive psychology. She received her doctorate degree in clinical psychology from Louisiana State University. She completed her doctoral internship at the Kennedy Krieger Institute through John Hopkins University in the Neurobehavioral Unit and continued this specialization through her postdoctoral fellowship in the Severe Behavior Program at the Marcus Autism Center through Emory University School of Medicine. Dr. Scheithauer is currently an associate professor in the Division of Autism and Related Disabilities in the Department of Pediatrics at Emory University's School of Medicine. She provides clinical care at the Marcus Autism Center, overseeing outpatient programs and managing cases in an intensive outpatient program. These programs use function-based treatments to target behaviors that challenge, such as aggression, self-injury, elopement, pica and disruption, exhibited by individuals with developmental and intellectual disabilities. In addition to providing direct clinical career, Dr. Scheithauer also trains doctoral interns, postdoctoral fellows and board-certified behavior analyst trainees.



Eva Schoeters, Chair, be-TSC, Co-Chair, TSC International (TSCi), Director, Rare Diseases Belgium, Mortsel, Belgium

Eva Schoeters is currently director at RaDiOrg, Rare Diseases Belgium. Her involvement in rare diseases originates from the diagnosis of her son, Pepijn (°2008), with TSC. Eva led the way to the establishment of the Belgian association for this disease, be-TSC, in 2015 and has been chair of the association since then. She became co-chair of TSCi in August 2018. She was, however, trained as an art historian and worked in the field of preservation of historical monuments for many years. Her work for TSC followed Eva's commitment to advocacy for rare diseases in general. After working at the sidelines as a volunteer in the Belgian rare disease organization, RaDiOrg, she became president in March 2018 and consecutively stepped out of this role to become professionally involved in 2019. It has become a mission to contribute actively to creating greater awareness and better care for all living with a rare disease, whether it be TSC or another one of the more than 6000 defined rare diseases.

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Rebecca J. Schultz, PhD, APRN, CPNP-PC, FAES, Associate Professor/Texas Woman's University and Assistant Professor/Baylor College of Medicine, Houston, TX

Rebecca J. Schultz received her PhD in Nursing Science from Texas Woman's University and Master of Science in Nursing from the University of Texas Galveston. She is an Associate Professor in the College of Nursing, Texas Woman's University, and an Assistant Professor/Pediatric Nurse Practitioner at Baylor College of Medicine/Texas Children's Hospital. Dr. Schultz has more than 30 years' experience as a Pediatric Nurse Practitioner, caring for youth with neurological disorders and their families. She is a strong advocate for individualized, family-centered care and acquisition of self-management skills to promote youth independence and successful transition to adulthood. Dr. Schultz lectures regularly at the local, state, national and international level on transition to adult care and management of seizures in the school. Her research interests include access to care, self-management skills, psychosocial impact of chronic illness on youth and family and transition from pediatric to adult health care for youth with neurological disorders. She serves on the Child Neurology Foundation Board of Directors and the Professional Advisory Board for the Epilepsy Foundation, Houston affiliate. She is an active member of the American Academy of Neurology Epilepsy Quality Measure Development Work Group, the Child Neurology Foundation Transition Project Advisory Committee and the Association of Child Neurology Nurses.



Julie Scroggins, TSC Alliance Volunteer, Burlington, WI

Julie has been dedicated to raising awareness and funding for TSC since her daughter Mary Ann's diagnosis. She founded Bcureful in 2011 and is very proud of all the organization accomplished and its continued impact today as the Bcureful Travel Fund at the TSC Alliance. She previously served on the TSC Alliance Board of Directors and has participated in the March on Capitol Hill for the past 13 years. Julie was the former Community Programs Manager, West for the TSC Alliance. In that role, one of her responsibilities was growing the Future Leader and Adult Regional Coordinator programs, which was her favorite part of the job.



Mary Ann Scroggins, TSC Adult, Grayslake, IL

After being diagnosed with TSC following a car accident in 2009, Mary Ann partnered with her mother, Julie, to establish a TSC nonprofit and has been advocating for TSC patients since. She recruits Research & Development talent in the pharmaceutical industry with the mission to improve patient's lives.



Susan Sherman, MHA, Executive Director & CEO The LAM Foundation, Cincinnati, OH

Susan Sherman, MHA, is the Executive Director & CEO of The LAM Foundation, a non-profit patient advocacy organization with a mission to urgently seek safe and effective treatments, and ultimately a cure, for lymphangioleiomyomatosis (LAM) through advocacy and the funding of promising research. Under Sue's eight years of leadership, the Foundation has distributed more than \$3.5 million in research funding to LAM investigators, hosted seven international research conferences and LAMposiums, implemented an annual in-person scientific advisory board study section and effectively integrated the patient voice into research design. Sue joined The LAM Foundation as Executive Director in 2013. She is a seasoned healthcare executive who has contributed to the advancement of health and the healthcare industry for more than 20 years. She holds a Master of Health Administration from Xavier University in Cincinnati, a Bachelor of Science in Biology from Indiana University and was formerly a certified executive coach. She has diverse industry expertise including healthcare administration, non-profit management, patient advocacy and business development.



Sean Shillinger, TSC Alliance Board of Directors, Adamstown, MD

Sean has a 7-year-old daughter, Brynleigh, who has TSC. Sean is the Population Health IT Supervisor at Frederick Health in Maryland, with expertise in population health, analytics, electronic health records, health information exchange, accountable care, practice transformation, and government health policy. He serves on the Science and Medical and Government Relations Committees and has participated on the Step Forward to Cure TSC National Walk on the Mall and DC SoundBites planning committees. Sean has been active in government advocacy efforts during the annual March on Capitol Hill and the Maryland Advocacy and Awareness Day. Sean and his wife Lauren also advocated for "Brynleigh's Act" recently passed into law making Maryland schools seizure safe.



Katie Smith, Director, Government & Global Affairs, TSC Alliance, Silver Spring, MD

Katie joined the TSC Alliance staff in 2006. She currently oversees the organization's international out-reach efforts, directs government advocacy efforts and co-chairs TSC International. During her tenure with the TSC Alliance, Katie has implemented the organization's Global Alliance program, which currently includes formal partnerships with Israel, Canada, Mexico, Thailand, India and Hungary.



Steven P. Sparagana, MD, Director, Pediatric Neurology and Rehabilitation Medicine, Scottish Rite for Children and Associate Professor, University of Texas Southwestern Medical Center, Dallas, TX

Steven P. Sparagana, MD, attained his medical degree at the University of Texas Health Science Center at San Antonio after completing his undergraduate and graduate degrees at Texas A&M University. He subsequently trained in pediatrics at St. Louis Children's Hospital, and in child neurology at Children's Health in Dallas as part of the neurology program at the University of Texas Southwestern Medical Center. Dr. Sparagana is a pediatric neurologist at Scottish Rite for Children where he is the Medical Director of Neurology and Rehabilitation Medicine and Director of the Tuberous Sclerosis Complex Clinic. He has special interests in TSC, cerebral palsy, and neurophysiology. He and his team are involved in clinical research on neurological matters (epilepsy, subependymal giant cell tumors, neuroimaging), and various aspects of renal angiomyolipomas (natural history, biopsies, and embolization) in children with TSC.



Kimberly Street, LMSW, LPN, CCM, Clinical Center for Adults with Neuro Developmental Disorders, Baltimore, MD

Kimberly Street was previously the Social Worker at the University of Maryland Clinical Center for Adults with Neuro-Developmental Disorders, which is a TSC Center of Excellence. Kimberly is also an adjunct professor at University of Maryland School of Social Work where she is an alum. Kimberly was the recipient of the gold standard in teaching recognition award for the academic year 2019-2020. In addition to her master's degree, Kimberly holds dual bachelor's degrees in social work and sociology, respectively. She is also a licensed practical nurse who was appointed to the Maryland State Board of Nursing by the governor where she served two terms, respectively. Kimberly has worked extensively in the community and managed 2 Licensed Assisted Living facilities in Baltimore County for 13 years. Prior to her role at UMB, she served as the Director of Support Services at Meals on Wheels of Central Maryland overseeing the internship and case management programs. Kimberly holds a certification in Case Management and co facilitated the didactics for the interprofessional education (IPE) program at the Presidents Clinic, an IPE initiative created by Dr. Perman, Chancellor of the University of Maryland. She currently serves on the TSC Alliance's Equity, Diversity and Inclusion (ED&I) Task Force. Kimberly also has an interest in family caregiving and blogs about her experience. In her spare time, she enjoys traveling and spending time with her family.

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Lisa Szilagyi, Director, The Aurelia Foundation-Creative Steps Adult Program, Los Angeles, CA

Lisa is a credentialed Special Education Teacher who has worked primarily with teens and young adults with moderate to severe disabilities. Lisa began her career in special education after the birth of her daughter, Emily. Emily was born in 1989 with TSC. Lisa worked at Malibu High school for more than 17 years teaching the Life Skills class and bringing a Person-Centered approach to working with students with disabilities. Lisa helped start the Best Buddies Club at Malibu High in 2002 and remained the Special Education advisor for the club through 2013. In 2010, Lisa collaborated with a group of parents to found the Aurelia Foundation, with the goal of creating a quality day program to serve the needs of adults with disabilities once they have left the school system. The Creative Steps Community Integration Program began in 2013 and currently supports more than 75 adults with a variety of different disabilities. Lisa is the Program Director for the Aurelia Foundation-Creative Steps Day program. The program has locations in Santa Monica, Westlake Village/Thousand Oaks, and our newest program location in Torrance. Lisa is also a Co-Founder and Co-Director of Hand in Hand, an after-school social skills inclusion program at the Malibu Jewish Center and Synagogue. In 2011, Lisa was honored to be a recipient of The Malibu Times Citizens of the Year Dolphin Award for her work with special needs students in the Malibu area.



Philip Tallman, TSC Alliance Adult Regional Coordinator, Aurora, IL

Phil Tallman resides in Aurora, IL with his wife and two children. He and his two children both have TSC. Phil works for an educational technology company as Vice President of Sales.



Joyce MC Teng, MD, PhD, Professor of Dermatology, Director, Pediatric Dermatology, Stanford School of Medicine, Palo Alto, CA

Dr. Teng is a professor of Dermatology and Pediatrics at Stanford University. She also serves as the chief of pediatric dermatology as well as the fellowship director. Prior to joining Stanford University, Dr. Teng was the chief of pediatric dermatology at the University of Wisconsin- Madison. Dr. Teng's has a special clinical and research interests in rare genetic skin disorders and directs the multidisciplinary genetic skin disorder clinic at Stanford Children's Health. Her translational research focuses on understanding the pathogenesis of genetic diseases and targeted therapy discovery. She is the chief editor of Pediatric Dermatology Therapy book, Therapy in Pediatric Dermatology textbook, In addition, she has authorized many book chapters as well as peer-reviewed articles. She published the first article in literature using topical sirolimus for the treatment of skin manifestations in TSC in 2010 and is the first author of published current quideline for the management of dermatologic manifestations in TSC.





Elizabeth Thiele, MD, PhD, Director, Pediatric Epilepsy Program and Director, Carol and James Herscot Center for Tuberous Sclerosis Complex, Massachusetts General Hospital, Boston, MA

Dr. Elizabeth A. Thiele is a neurologist and epileptologist at Massachusetts General Hospital. She received her medical training at Johns Hopkins University School of Medicine in Baltimore, Maryland, and completed an internship and residency in pediatrics at the Johns Hopkins Hospital. She completed a second residency in child neurology and a postdoctoral research fellowship in neurology at Children's Hospital in Boston. Dr. Thiele organized and established the Herscot Center for Tuberous Sclerosis Complex, a multidisciplinary comprehensive clinical program for TSC, as well as a ketogenic diet clinic to treat and manage patients with epilepsy. She is also the Director of the Pediatric Epilepsy Service at Mass General and a Professor in Neurology at Harvard Medical School. Dr. Thiele's research and clinical interests include the role of diet in epilepsy treatment, genotype-phenotype correlation in TSC, the role of epilepsy surgery in management of intractable epilepsy, outcomes following infantile spasms, and neuropsychological profiles in relationship to tuber number and location in TSC.



Nadia D. Vann, Attorney, Lansing, MI

Nadia D. Vann is an attorney practicing in Michigan. Her practice areas include estate planning and administration and special needs planning for families. Nadia has practiced in the area of education law, focusing on special education law and advocacy. Before entering the legal profession, Nadia worked as a special education teacher and teacher consultant in school districts in southeast Michigan. Nadia has trained attorneys, advocates and educators in best practices for maintaining harmony among families through proper and succinct estate planning and planning for disabled children and adults throughout their lives. Nadia currently serves as chair of the Disability Rights Committee of the Elder Law and Disability Rights Section of the State Bar of Michigan, where she is a Council member. She also serves on the boards of the Epilepsy Foundation of Michigan and MiACRES, a special education advocacy non-profit.



Vanessa Y. Vazquez, TSC Parent, Miami, FL

Vanessa's son, Max Lucca, was diagnosed with TSC in 2008, just two weeks after being born. She immediately turned to the TSC Alliance for help, guidance and support. Shortly after, her family hosted their first Step Forward to Cure TSC walk and has been involved ever since. Max Lucca is an amazing kid and he inspires his family every day to advocate for those affected, to support those who most need it and to ensure our Spanish-speaking community has access to services. Vanessa is honored to serve as Co-Chair for the TSC Alliance of Florida and serves on the TSC Alliance Board of Directors and the Equity, Diversity and Inclusion (ED&I) Task Force.



Zacil Vilchis, MD, MsC, Clinical Geneticist, Research Assistant Professor, TSC Alliance of Mexico, Mérida, México

Dr. Zacil Vilchis is a Specialist in Medical Genetics who graduated from the Autonomous University of Mexico and earned a Master's degree in biomedical sciences from the Autonomous University of Yucatan. Dr. Vilchis is a full-time Research Associate Professor affiliated to the Human Genetics Laboratory of the Biomedics Research Center of the Autonomous University of Yucatán since 2018 and active member of the Mexican Association of Human Genetics. She's been involved with multiple research projects, among which are the analysis of genomic variants in breast cancer, the genetic analysis of patients with congenital deafness and is a national expert in TSC. Highlights of her community work include being a Founding Member and Co-Director of the first TSC clinic in Mexico, active since 2016 and a member of the Medical Council of the Mexican Association of TSC (AMET) and Senior Advisory Board of the TSC Alliance of Mexico. She also serves on the medical council of the Yucatecan Pro-Hearing Impaired Association and collaborator of the Yucatan Project which, in collaboration with the Universities of Florida and Oklahoma, supports families with congenital deafness since 2014.

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Kristi Viles, PhD, Director Alliance Management, BridgeBio Gene Therapy, Raleigh, NC

Kristi Viles is the Director of Alliance Management for BridgeBio Gene Therapy (BBGT). Kristi leads program management and cross-functional and external scientific alignment activities of the early-stage gene therapy programs. She guides and facilitates operational execution of the early-stage pipeline. Kristi is passionate about developing novel gene therapies for diseases with significant unmet need. Previously, she was the Senior Program Manager for Precision BioSciences and a Clinical Trial Project Leader for Duke Translational Research institute. Kristi has a PhD in Genetics and Genomics from Duke University. The BBGT team is working with the TSC Alliance and leaders in the TSC field to develop our TSC gene therapy to bring it to the clinic and the TSC community.



Rahul Vipparthi, Managing Trustee, TSC Alliance of India, Madhapur, Hyderabad

Rahul Vipparthi is the Managing Trustee of TSC Alliance of India. Rahul's 14-year-old daughter was diagnosed with TSC in 2007. He is working to help families better manage TSC by building the capacity of the TSC community and raising awareness of TSC clinical practices through engaging the various stakeholders. Rahul's passion is in promoting TSC research in Indian Institutions. Rahul has a strong background in Information Technology and Management.



Valerie M. Volkert, PhD, BCBA-D, Site Manager, Children's Multidisciplinary Feeding Program at the Marcus Autism Center, Atlanta, GA

Valerie M. Volkert, PhD, BCBA-D, Associate Professor (Pediatrics), is a psychologist site manager in the Children's Multidisciplinary Feeding Program at Marcus Autism Center. She received her doctorate in school psychology from Louisiana State University in 2007. She was previously faculty at the Munroe-Meyer Institute and adjunct faculty at the University of Nebraska at Omaha, where she became the Training Director for the Applied Behavior Analysis PhD Program and Center for Autism Spectrum Disorders. In 2015, Dr. Volkert joined the Marcus Autism Center. She sees patients in the intensive day treatment and outpatient clinics of the Feeding Program, supervises interns and residents and pursues lines of clinical research. Of particular interest are treatments to increase advanced feeding skills (e.g., chewing), synthesizing the findings of studies on the treatment of pediatric feeding disorders and evaluating alternative approaches to the treatment of chronic feeding problems (e.g., sensory integration and tube weaning). She has authored four book chapters and published 40 peer-reviewed research studies in multiple journals, including the Journal of Applied Behavior Analysis, the Journal of Pediatrics, Research in Developmental Disabilities, Behavior Modification, and Behavior Analysis in Practice. Dr. Volkert served as the Editorial Assistant for the Journal of Applied Behavior Analysis from 2008 to 2010. She serves on the board of editors for two journals and as associate editor for the Journal of Applied Behavior Analysis.



Oded Volovelsky, MD, PhD, Pediatric Nephrology Unit Head, Hadassah Hebrew University Medical Center, Jerusalem, Israel

Dr. Volovelsky feels privileged to treat patients with TSC and look for novel interventions to alleviate their disease. He is currently the head of the pediatric nephrology unit and research lab at Hadassah Hebrew University Medical Center in Jerusalem, Israel. He was trained as MD-PhD and pediatric resident at Hadassah and as a pediatric nephrology clinical fellow and post-doc student at Cincinnati Children's Hospital Medical Center in Ohio. Dr. Volovelsky's research goal is to improve kidney disease care of children and adults with TSC by revealing novel molecular pathways that control their kidney disease. His professional skill set includes an extensive study of the TSC-mTOR pathway in different organs and disease states and rich clinical experience in managing kidney disease in pediatric TSC patients. His research interests in this field include the mechanisms of TSC cystic kidney disease and the role of TSC in developing kidney and parathyroids of patients with kidney disease. In addition to establishing a research lab in the TSC field, Dr. Volovelsky co-founded a multidisciplinary unit for pediatric and adult patients with TSC from the region.



Howard L. Weiner, MD, FACS, FAAP, FAANS, Chief of Neurosurgery and George A. Peterkin Jr. Endowed Chair in Neurosurgery, Texas Children's Hospital, Professor and Vice Chair of Neurosurgery, Baylor College of Medicine, Houston, TX

Howard L. Weiner, MD, is Chief of Neurosurgery and the George A. Peterkin Jr. Endowed Chair at Texas Children's Hospital, where he leads a team of six pediatric neurosurgeons and is Professor and Vice Chair of Neurosurgery at Baylor College of Medicine in Houston. Prior to this, he was Professor of Neurosurgery at NYU Langone Medical Center, where he worked for 27 years. He received his BA from the University of Pennsylvania and MD from Cornell University Medical College, graduating from both with the highest academic standing in his class. He completed neurosurgery residency and pediatric neurosurgery fellowship at NYU and was the AANS Van Wagenen Fellow in the lab of Nicole Le Douarin, PhD, in Paris. He conducted basic brain tumor research in the laboratory of HHMI Investigator and Professor Ed Ziff at NYU and was awarded an NIH grant. Howard has developed an interest in pediatric epilepsy and brain tumor surgery and is considered an expert in the treatment of TSC. He has served on the Editorial Board of JNS: Pediatrics and Childs Nervous System, completed a term on the Executive Council of the American Society of Pediatric Neurosurgeons, serves as an Examiner for the American Board of Neurological Surgeons and is a member of the Society of Neurological Surgeons and American Academy of Neurological Surgery.



Klaus Werner, MD, PhD, Director, TSC Center of Excellence, Duke University Medical Center, Durham, NC

Dr. Werner is a Pediatric Neurologist with expertise and interest in pediatric headaches and TSC. He has a specific interest in cognitive processing/autism in TSC and the continuous need/care of adolescents/adult patients with TSC. He studied medicine in Germany and at the Pediatric Hospital Robert Debre in Paris and earned his MD from the Institute of Pharmacology, University of Bonn with the thesis title being "Characterization of the pre-synaptic inhibitory serotonin (5-HT) receptor of the saphenous vein." Dr. Werner's pediatric training was in London and Cambridge (UK) followed by training in child neurology at the Hospital for Sick Children in Toronto. He earned his PhD from the Institute of Child Health, University College London with the title "Auditory processing in the syndrome of infantile spasms." He has been in practice as a pediatric neurologist since 2012 and been on faculty at Duke University since 2016. Dr. Werner is currently the Director of the Pediatric Headache/Botox Clinic and Multi-Disciplinary TSC Center of Excellence for children and adults at Duke University. He has served on the Professional Advisory Board for the TSC Alliance since 2018. Dr. Werner has been a member of the TSC Clinical Research Consortium since 2018 and TSC Clinic Committee since 2021.



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James W. Wheless, BScPharm, MD, FAAP, FACP, FAAN, FAES Professor & Chief of Pediatric Neurology, Le Bonheur Chair of Pediatric Neurology, University of Tennessee Health Science Center, Director, Le Bonheur Comprehensive Epilepsy Program & Neuroscience Institute, Co-Director, Le Bonheur Tuberous Sclerosis Complex Center of Excellence, Le Bonheur Children's Hospital, Memphis, TN

Dr. James Wheless is an experienced neurologist and researcher, whose research is focused on pediatric anti-epileptic drug development, the ketogenic diet, epilepsy surgery and magnetoencephalography. Dr. Wheless is the Professor and Chief of Pediatric Neurology and the Le Bonheur Chair in Pediatric Neurology at the University of Tennessee Health Science Center in Memphis. He also serves as Director of the Neuroscience Institute and the Le Bonheur Comprehensive Epilepsy Program for the Le Bonheur Children's Hospital. Dr. Wheless is also an Adjunct Clinical Faculty Member in the Department of Pediatric Medicine at St. Jude Children's Research Hospital. He is a Diplomat of the American Board of Pediatrics and the American Board of Psychiatry and Neurology with special qualifications in Child Neurology, Clinical Neurophysiology and Epilepsy. Dr. Wheless is a Fellow of the American Academy of Pediatrics, the American Academy of Neurology and the American Epilepsy Society. He is a member of the Editorial Board of Journal of Child Neurology, Formulary and Epilepsy.com and serves as reviewer of a number of journals including Neurology, Epilepsia, Pediatrics and Epilepsy and Behavior. His primary interests include childhood convulsive disorders. He has authored more than 800 chapters, articles and abstracts on these subjects and is the editor of four textbooks on epilepsy. Dr. Wheless lectures widely on pediatric epilepsy. He oversees an interdisciplinary team of pediatric specialists that treat epilepsy patients with the most effective medical treatments available.



Michael Wong, MD, PhD, Allen P. and Josephine B. Green Professor of Pediatric Neurology, Washington University School of Medicine, St. Louis, MO

Michael Wong, MD, PhD, is the Allen P. and Josephine B. Green Professor of Pediatric Neurology at Washington University School of Medicine. He received a BA in Biology from Princeton University and MD and PhD (neuroscience) degrees from University of Texas Southwestern Medical School. He then moved to Washington University in St. Louis for pediatric neurology residency and epilepsy fellowship, before becoming a faculty member in the Department of Neurology. He is a board-certified pediatric epileptologist and clinical neurophysiologist. He is also director of the Washington University Tuberous Sclerosis Center, which provides multidisciplinary care for children and adults with TSC and integrates both basic science and clinical research in TSC. Dr. Wong's basic science laboratory primarily investigates mechanisms of epileptogenesis and seizure-induced brain injury in genetic and acquired rodent models of epilepsy. His laboratory has been among the leaders in establishing the critical role of the mechanistic target of rapamycin (mTOR) pathway in epileptogenesis in TSC, as well as in animal models of acquired epilepsy due to brain injury. Directly related to his basic research, he has served as site principal investigator at Washington University for a number of clinical trials, such as the EXIST-3 trial of the mTOR inhibitor everolimus for epilepsy in TSC (leading to its FDA approval for that indication) and the PREVeNT trial for preventing neurological manifestations of TSC. The latter trial is particularly impactful in attempting to develop a first preventative anti-epileptogenic drug treatment for epilepsy. He has been funded continuously by NIH and other sources for 20 years and has co-authored more than 100 basic and clinical research papers and reviews. He is past recipient of the American Academy of Neurology Dreifuss-Penry Epilepsy Research Award and is currently an Associate Editor of Epilepsia Open and Chief Editor of Epilepsy Currents.

Exhibit Hall and TSC Resource Center

The conference Exhibit Hall and TSC Resource Center, located in Grand Ballroom C, feature a wide array of service providers and vendors who will provide information, products and services most helpful to the TSC community.

Friday, July 29 8 a.m. to 5 p.m.

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The LAM Foundation urgently seeks safe and effective treatments, and ultimately a cure, for lymphangioleiomyomatosis (LAM) through advocacy and the funding of promising research. We are dedicated to serving the scientific, medical and patient communities by offering information, resources and a worldwide network of hope and support.

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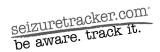


Seattle Children's, formerly Children's Hospital and Regional Medical Center, formerly Children's Orthopedic Hospital, is a children's hospital in the Laurelhurst neighborhood of Seattle, Washington. The hospital specializes in the care of infants, children, teens, and young adults aged 0–21 in several specialties.

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Upsher-Smith Laboratories, LLC is a trusted U.S. pharmaceutical company that has strived to deliver quality, affordable generic medications for nearly a century. In June 2017, Upsher-Smith was acquired by Sawai Pharmaceutical Co., Ltd., a large publicly traded generics company in Japan that had been seeking entry into the U.S. market. Upsher-Smith and Sawai share a strikingly similar family history and hold many of the same cherished goals and valuesmost importantly, the philosophy of always putting patients first. Upsher-Smith will continue to do those things it does best, which is provide a consistent supply of quality, affordable medications and invest in its historically strong industry relationships. Ultimately, Upsher-Smith believes the acquisition by Sawai represents a tremendous opportunity to leverage each company for growth worldwide and embark together on an exciting new chapter in generics.

Glossary

Organizational Acronyms

CDC: Centers for Disease Control & Prevention

CDMRP: Congressionally Directed Medical Research Program

DEA: Drug Enforcement Administration

DoD: Department of Defense

EMU: European Medicines Agency (EMU)

HHS or DHHS: Department of Health and Human Services

FDA: Food and Drug Administration IRB: Institutional Review Board NIH: National Institutes of Health ORDR: Office of Rare Disease Research SSI: Supplemental Security Income

TSCRP: Tuberous Sclerosis Complex Research Program

WHO: World Health Organization

Medical Acronyms

ABA: applied behavioral therapy **ACTH:** adrenocorticotropic hormone

ADHD: attention-deficit hyperactivity disorder

AML: angiomyolipoma

ASD: autism spectrum disorder

BCBA: board-certified behavioral analyst

CBD: cannabidiol

CNS: central nervous system CBT: cognitive behavioral therapy

CT (or CAT scan): computerized tomography

Echo: echocardiogram

EKG (or ECG): electrocardiogram EEG: electroencephalogram GABA: gamma-aminobutyric acid GFR: glomerular filtration rate

HR CT: high-resolution computed tomography

MMPH: multifocal micronodular pneumocyte hyperplasia

NMI: no mutation identified

OB-GYN: gynecology; obstetrics and gynecology **PanNET (or PNET):** pancreatic neuroendocrine tumor

PET: positron emission tomography **PFDD:** patient-focused drug development

PKD: polycystic kidney disease **PNS:** peripheral nervous system

SEGA: subependymal giant cell astrocytoma **TAND:** TSC-associated neuropsychiatric disorders

TSC: tuberous sclerosis complex VNS: vagus nerve stimulator

VUS: variant of unknown significance

acute

Abrupt onset and/or of short duration. Contrast with chronic.

adrenocorticotropic hormone (ACTH)

ACTH is a hormone that is important during the body's reaction to stress. It can be used as a second-line therapy for infantile spasms in TSC if vigabatrin treatment is unsuccessful.

adverse effects; side effects

An adverse effect is an undesired, often harmful, effect resulting directly from a medication/intervention.

advocacy

Activity that aims to influence decisions within political, economic, and social systems/institutions.

allele

One copy of a gene. Individuals with TSC generally have one normal allele and one disease-causing allele.

aphthous ulcers (canker sores)

Small, shallow lesions that may form in the mouth, sometimes as a side-effect of mTOR inhibition therapy.

applied behavioral analysis (ABA) therapy

Applied behavioral analysis (ABA) is a type of therapy that can improve social, communication, and learning skills through reinforcement strategies. It is sometimes used as an intervention to treat autism.

angiofibroma

A nonmalignant tumor composed of blood vessels and fibrous tissue typically on the face. They initially appear as pink or red bumps and can form a butterfly-shaped distribution around the nose, cheeks, and chin.

angiomyolipoma (AML)

Angiomyolipomas are nonmalignant tumors mainly composed of blood vessels and fatty tissue. In TSC they are most commonly found in the kidneys but can also be found in the lungs of individuals with lymphangioleiomyomatosis (LAM). Please note: In other medical contexts outside of TSC, the acronym "AML" may refer to acute myeloid leukemia, a type of malignant blood cancer.

anti-seizure medication (ASM); anti-epileptic drug (AED)

Any drug that is prescribed to treat or prevent seizures or epilepsy. Previously, the term "anti-epileptic drug" (AED) was used more commonly; current practice is to use the term "anti-seizure medication" (ASM) to be more inclusive.

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animal model

A non-human animal (such as a mouse, rat, or zebrafish), often genetically modified to mimic human gene mutations, used to research human disease.

astrocvte

A particular type of glial cell (a type of non-neuron brain cell) that is star shaped.

attention-deficit hyperactivity disorder (ADHD)

ADHD is generally considered to be a developmental disorder characterized by persistent inattention and/or hyperactivity-impulsivity.

autism spectrum disorder (ASD)

Complex brain disorder characterized by problems with social communication, insistence of sameness of activities and surroundings, and repetitive patterns of behavior.

autosomal dominant

An "autosome" is one of the 22 non-sex (X or Y) chromosomes in the human genome. Almost all cells in the body contain two copies, or alleles, of each of the 22 autosomes. If a disorder follows an autosomal dominant inheritance pattern, it means only one allele needs to be mutated to cause said disorder. An affected parent has a 50% chance of passing the disorder to his or her offspring.

basic research

Basic research is necessary laboratory research that provides a foundation of knowledge. Typically, basic science research focuses on determining the causal mechanisms behind the functioning of the human body in health and illness.

benign tumor

A mass of cells that does not invade surrounding tissue and usually do not grow back if completely removed. They typically grow more slowly than malignant tumors. Although not cancerous, they can cause symptoms and, in some cases, may be life-threatening.

biobank; biorepository

A large collection of biological samples or medical data collected for distribution for research purposes.

biochemical pathway

A linked series of chemical reactions driving by enzyme within a cell.

biomarker

A biomarker is any measurable substance (such as blood sugar) or characteristic (such as blood pressure) that may be indicative of some biological event, disease, or response to a certain environment or treatment.

biospecimen or biosample

A biological sample donated by a patient and collected by a biorepository for use in research.

blood-brain barrier

The blood-brain barrier is a network of blood vessels and tissue that surrounds the brain. The barrier is permeable to certain substances, such as water, oxygen, carbon dioxide, and some general anesthetics, but it is not permeable to bacteria and many types of drugs.

brand-name drug

A drug produced by the original manufacturer who brought the product through the original country's official approval process and sold under its trade name. Contrast with generic drug.

buccal swab; cheek swab

A process of swabbing the insides of one's mouth to collect cheek cells for DNA analysis.

cannabidiol (CBD)

A specific chemical component occurring naturally in the cannabis (hemp or marijuana) plant that can be isolated from cannabis or synthesized in a laboratory. A specific formulation of CBD (Epidiolex®) was approved by the FDA in 2020 for treatment of seizures associated with TSC.

cannabis; cannabinoid

Cannabis is both a general term for products of the hemp or marijuana plant Cannabis sativa L. and a genus including the species: Cannabis sativa L. and Cannabis indica. A cannabinoid is a chemical molecule naturally occurring in the cannabis plant.

cardiac rhabdomyoma

A nonmalignant tumor composed of muscle tissue that occurs in the heart.

central nervous system (CNS)

The CNS consists of the brain and spinal cord.

cephalic fibrous plaque

Previously known as a "forehead fibrous plaque," the cephalic fibrous plaque is similar to an angiofibroma but is a larger area of elevated, pink skin typically found on the forehead, cheeks or scalp.

chromosome

Chromosomes are tightly coiled packages of DNA (deoxyribonucleic acid). Each human has 23 unique pairs of chromosomes, 22 of which are called "autosomes" or "autosomal chromosomes." The 23rd pair of chromosomes is sex-defining and consists of two X chromosomes in females or one X and one Y chromosome in males.

A condition is chronic if it is long-lasting and does not spontaneously resolve. Contrast with acute.

chyle or chylous effusion

See: pleural effusion; chylothorax

circadian rhythm

The mental, physical, and behavioral changes that follow a daily (24-hr.) cycle. These changes respond primarily to light/darkness—e.g., sleeping at night and being awake during the day is a light-related circadian rhythm.

clinical trial

A study that assigns participants to one or more interventions to determine whether the intervention has an impact on health outcomes.

Phase 1 clinical trial: Also called a "first-in-human study," a Phase I clinical trial tests a new drug in humans to see if it is safe to use. This is typically done in a small number of healthy volunteers.

Phase 2 clinical trial: Tests whether a new drug is at all effective in humans. This is typically done in a small number of people with the disease. There is usually a group of participants who receive a placebo.

Phase 3 clinical trial: Tests how safe and effective the new drug is in a larger group of people with the disease and usually includes a group of participants who receive a placebo. One or more Phase 3 trials are generally required for FDA approval.

Phase 4 clinical trial: Phase 4 studies may be required by FDA after approving a new drug to identify and evaluate the long-term effects with a larger patient population.

Clinical Consensus Guidelines for Diagnosis, Surveillance and Management for TSC

Most recently updated in 2021, the Updated International Tuberous Sclerosis Complex Diagnostic Criteria and Surveillance and Management Recommendations are a peer-reviewed set of recommendations compiled by international TSC experts that outline the internationally recognized, evidence-based clinical practices for TSC. These guidelines are seen as international gold standards for TSC care.

cognition

Cognition refers to the mental process of thinking, learning, remembering, being aware of surroundings and using judgment.

cognitive behavioral therapy (CBT)

CBT is a type of psychotherapy that focuses on talking with a mental health professional to investigate current problems and their possible solutions. CBT can sometimes involve "homework," which might include journaling, performing certain thought exercises or other techniques a therapist might suggest.

computerized tomography (CT): CAT scan

Medical imaging using a radioactive tracer to create cross-sectional images of a patient that allow medical professionals to see internal organs and their contents without cutting. Sometimes called "CAT" scans for "computed axial tomography" or "computer aided tomography."

control group

A control group in an experiment or study does not receive treatment but may receive placebo and is used as a benchmark or comparison point for the study intervention.

controlled substance (see also Schedules I-V)

A chemical or drug whose possession, manufacture, or use is regulated by the government. In the United States, controlled substances are rated as Schedule I, II, III, IV, or V by the Drug Enforcement Administration (DEA).

coordinated care

The coordination of health care across multiple disciplines and specialties; often includes a multidisciplinary team.

cortical tuber

An area of the brain that contains a disorganized collection of abnormal cells usually found in the outer layers of the brain (called the "cortex") in people with TSC but can also be found in deeper areas of the brain.

cyst

A closed sac containing fluid or semisolid material, developing abnormally in a body cavity or structure. Cysts can impair the function of surrounding tissue.

dental pits

Pits or small areas of decay in the coating protective enamel coating of teeth. Not all dental pits are cavities, but food may build up in these pits and later lead to a cavity.

dermatology

Dermatology is the study of the hair, skin, nails and the diseases that involve them.

developmental delay

Delay in the normal cognitive and/or physical development of a child.

developmental medicine

Developmental medicine (often paired with child neurology) concerns itself with improving outcomes of children facing developmental and behavioral challenges.

double-blind clinical trial

A study in which both the investigator and the participant are blind to (unaware of) whether the participant is in the control group or receiving active treatment.

Drosophila

A genus of flies belonging to the "small fruit fly" family (Drosophilidae). The species D. melanogaster is particularly important to genetic research and is considered an animal model.

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echocardiogram (echo)

A method for imaging the heart using ultrasound (sonogram) technology.

Eker rat

The Eker rat was one of the earliest animal models for TSC; it has a naturally occurring heterozygous mutation in TSC2. While they do not have spontaneous epilepsy, they do appear to have a mild epilepsy phenotype. The Eker rat spontaneously develops tumor-like lesions on their kidneys.

electrocardiogram (EKG or ECG)

A method for monitoring the heart's electrical rhythm by using ten electrodes placed on a patient's skin.

electroencephalogram (EEG)

An EEG uses surface electrodes placed on a patient to monitor the brain's electric activity. Sometimes it is helpful to record a video in addition to performing an EEG to further investigate the type and severity of seizure recorded by an EEG.

eligibility criteria

Characteristics required of people who are allowed to participate in a particular clinical study, such as age requirements, seizure frequency, etc.

endocrinology

Study of the endocrine system (such as pituitary, thyroid, and adrenal glands as well as the pancreas, ovaries, and testes) and hormones.

enzyme

An enzyme is a biological catalyst, usually a type of protein, that helps facilitate biochemical reactions.

epilepsy

When a person has two or more seizures that have not been provoked by specific events such as trauma, infection, fever or chemical change, he or she is considered to have epilepsy.

epileptology

Study of epileptic seizures and seizure disorders, anticonvulsant therapies and monitoring.

everolimus (Afinitor® in the US; Zortress®, Votubia®, or Certican® elsewhere)

Everolimus is an mTOR inhibitor used to treat a variety of conditions. In TSC, it is used to treat SEGAs that cannot be removed surgically, renal angiomyolipomas and partial-onset seizures.

formulary (also called "preferred")

A list of prescription drugs covered by a prescription drug plan or another insurance plan or national health system offering prescription drug benefits. Prescription drugs included on the formulary may also be referred to as "preferred" drugs; whereas those not included on the formulary are called non-formulary or "non-preferred" drugs.

gamma-aminobutyric acid (GABA)

GABA is the main inhibitory neurotransmitter—meaning, it plays an important role in lowering neuronal excitation. Some seizures are treated by increasing GABA's effectiveness: vigabatrin works by decreasing the breakdown of GABA.

gastroenterology

Gastroenterology is the study of the normal function and diseases of the esophagus, stomach, small intestine, colon and rectum, pancreas, gallbladder, bile ducts and liver.

A portion of a chromosome used by a cell to guide the creation of a protein.

generic drug

A version of a drug marketed after the original (brandname) manufacturer no longer has exclusive rights to market a drug. Generic drugs must demonstrate to FDA they release the same amount of active ingredient as the brand-name drug.

genetic counselor

Genetic counselors are professionals who have specialized education in genetics and counseling to provide personalized help an individual or family may need as they make decisions about their genetic health.

genetic disorder

A genetic disorder is a disorder that is caused by one or more abnormalities in a gene or chromosome present from birth. They may be hereditary (passed down from parents' genes) or they can be spontaneous.

Genotype refers to the sequence of DNA code corresponding to a person's alleles of a certain gene.

The combining of an egg and sperm cell to create a viable embryo is called "germination." DNA contributed by both the mother's egg and the father's sperm cell is "germline" DNA and becomes the source of DNA for all other cells in the child's body.

gingival fibromas

Fibrous nodular growths involving the gingiva (gums) in the mouth.

glomerular filtration rate (GFR)

Rate at which the kidneys filter fluid, an important measurement of renal function.

grey matter

Greyish pink tissue in the brain, which is primarily made up of neuronal cell bodies.

gynecology; obstetrics and gynecology (OB-GYN)

Gynecology is the medical practice dealing with the health of female reproductive systems (vagina, uterus, ovaries) and the breasts.

hamartin

Hamartin is the protein encoded by the TSC1 gene. With the product of TSC2, tuberin, it is a key component of the TSC protein complex, a group of proteins inside a cell that, together, regulate mTORC1 activity.

hamartoma

A benign tumor in an organ composed of tissue elements normally found at that site but growing in an abnormal way. high-resolution computed tomography (HR CT)

A high-resolution CT scan is very similar to a normal CT scan, though used more often in diagnosis and investigation of the lungs.

hormone

The body's chemical messengers released into the bloodstream by endocrine organs (i.e., pancreas, thyroid gland, testes, etc.) to affect processes like growth, metabolism, sleep and mood.

hypomelanotic macule

Patch of skin with less color (hypo = less, melanin = pigment found in skin) than normal and is commonly found in TSC. These spots are benign and pose no physical threat.

hypsarrhythmia

Hypsarrhythmia is disorganized, chaotic pattern of brain waves that often accompanies infantile spasms.

immunosuppression

Suppression of the body's immune system and its ability to fight infections/other diseases. It may be deliberately induced with drugs (e.g., after an organ transplant) or it may result from certain diseases.

infantile spasms

A type of seizure that typically occurs between the ages of 2 and 24 months. It is identified by sudden jerks caused by muscle contractions, flexing of the body and neck and stiffening of the limbs. Each of these seizures lasts a very short time but can occur in long or short clusters. May be referred to by its new classification as either focal, generalized or unknown onset epileptic spasms.

informed consent

The process by which a person learns about the purpose, benefits and potential risks of a medical or surgical intervention or a research study and then agrees to receive the intervention or participate in the study.

interventional radiology

A sub-specialty of radiology using radiographic imaging methods to facilitate minimally invasive treatment, for example, when treating angiomyolipomas with embolization.

intractable epilepsy

If a patient's seizures do not respond well, or at all, after attempting multiple anti-epileptic treatments. Sometimes also called "refractory" or "uncontrolled" epilepsy.

Institutional Review Board (IRB)

A group of people who review, approve, and monitor the protocol of a research study involving human subjects. Their role is to protect the rights and welfare of people participating in a study.

ketogenic diet

A diet consisting of high-fat, low-carbohydrate foods typically prescribed by a physician and monitored by a dietician as adjunctive therapy for seizures that have not responded to other therapies.

knockout; knockout mouse

An organism, usually a mouse, having a certain genetic sequence removed, or "knocked out." Many animal models are created by knocking out a gene or part of a gene.

laser ablation

A technique using a thin laser probe to heat tissue to kill or remove ("ablate") tumors or lesions, often in the brain. Positioning of the probe and monitoring of heat is often performed using MRI.

lymphangioleiomyomatosis (LAM)

LAM is a lung disease caused by mutations in the TSC1 or TSC2 genes. It is characterized by an abnormal growth of smooth muscle cells, especially in the lungs, lymphatic system and kidneys that primarily occurs in women. Unregulated growth of these cells can lead to loss of lung function, accumulation of lymph rich-fluid in the chest and abdomen and growth of tumors in the kidneys. A person can have LAM without meeting clinical criteria for a TSC diagnosis.

magnetic resonance imaging (MRI)

MRI technology makes use of the hydrogen found abundantly within a patient's tissues (namely, fat and water). The tube-like machine surrounds the patient with a powerful magnet to measure subtle changes induced in the body by the magnetic field, enabling experts to visualize structures inside the body at very high resolution.

malignant tumor

A malignant tumor is cancerous. The primary hallmarks of malignant tumors are unregulated cell growth, stimulation of new blood vessel growth to feed the tumor and invasion of tissues, or metastasis.

metastasis

The abnormal spread of cells or a tumor from an original location to a new site within the body.

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mosaicism

When a mutation in a gene is present in some, but not all, cells in the body.

mTOR inhibitors

A class of drugs that inhibit the "mechanistic target of rapamycin" (mTOR), such as rapamycin (also known as sirolimus) and everolimus.

mTOR pathway

The "mechanistic target of rapamycin" (mTOR) is an enzyme regulating cellular metabolism and growth. The mTOR pathway is comprised of dozens, and maybe hundreds, of proteins that directly or indirectly regulate mTOR's function, or that are themselves directly or indirectly regulated by mTOR's function.

multifocal micronodular pneumocyte hyperplasia (MMPH) MMPH consists of an overgrowth (hyperplasia) of pneumocytes (a specific cell type in the lung). MMPH typically does not produce clinical symptoms but is often seen in TSC and may be present with or without LAM.

mutation

A change in the structure of a gene, whether a single base change in DNA or a deletion, insertion or rearrangement of a larger section of DNA.

natural history database (TSC Alliance Natural History Database and ToSCA)

A systematic compilation of information about how a disease affects different individuals over time. The TSC Alliance Natural History Database and the Tuberous SClerosis registry to increase disease Awareness (ToSCA) are two examples of multicenter, international disease registries to assess manifestations, interventions and outcomes in patients with TSC.

nephrology

Study of the kidneys, including diseases of the kidneys and normal kidney functioning.

neuro-oncology

Neuro-oncology is the study of tumors or cancer in the brain and spinal cord.

neurology

Neurology is the study of the nervous system, including the brain, spinal cord and peripheral nerves.

An electrically active cell that communicates information to other cells. The brain contains the highest concentration of neurons, but neurons extend from the spinal cord into every part of the body.

neurotransmitter

A chemical messenger released by a neuron used to communicate with other cells. Different neurotransmitters have different effects depending on the receptor protein with which the neurotransmitter interacts.

newborn screening

A process for testing all infants shortly after birth for a defined set of disorders that benefit from early treatment before the disorder is likely to be detected clinically. Newborn screening programs are often run by regional or national governments who define the set of disorders to be tested.

no mutation identified (NMI)

A situation in which a person meets the clinical criteria for diagnosis of a genetic disorder, but genetic testing fails to detect a disease-causing mutation. This may be due to mosaicism or the presence of mutations in a region of DNA not included in the genetic test.

occupational therapy

Therapy to help a person recover or develop skills needed for the activities of daily life. It can help develop, improve, sustain or restore independence.

ophthalmologists; optometrists; opticians

Ophthalmologists are physicians who are experts in diseases of the eye. Optometrists are licensed to conduct eye exams, detect certain abnormalities and prescribe contact lenses and glasses. Opticians are medical technicians who are trained to design, verify and fit eyeglass lenses, frames or contact lenses.

orphan drug

A medicinal product intended to treat a rare medical condition, or orphan disease. An "orphan disease" is a rare disease for which the population is so small that treatments may not be developed by the pharmaceutical industry for economic reasons.

pancreatic neuroendocrine tumor (PanNET; PNET)

Neuroendocrine tumors arise from cells of the endocrine (hormonal) and nervous system. While many are benign, some are malignant. Pancreatic NETs are different than the usual form of pancreatic cancer.

pathogenic mutation; pathogenic variant

A mutation in DNA known to cause a genetic disorder. In TSC, more than 1,500 unique pathogenic mutations have been found, but all lead to loss of function of TSC1 or TSC2 proteins.

pathology

Pathology is the study of disease. Clinical pathology is a medical specialty concerned with the diagnosis of disease based on laboratory analysis of bodily fluids and tissues.

patient-focused drug development (PFDD)

PFDD is a program to ensure that patients' experiences, perspectives, needs and priorities are captured and meaningfully incorporated into drug development and evaluation by FDA. The TSC Alliance sponsored a PFDD meeting in June 2017. A Voice of the Patient Report, a detailed summary of this meeting, was submitted to the FDA and is available on our website.

peripheral nervous system (PNS)

Comprised of neurons extending beyond the CNS, including sensory neurons for touch and pain, and motor neurons which regulate muscle function.

pharmacy drug list (PDL)

A list of prescription drugs that your specific insurance plan covers. This can be found in your plan terms.

phenotype

The characteristics associated with a person's condition. Two people can have the same phenotype but different genotypes, or the same genotype but different phenotypes. Both of these situations can be observed in TSC.

placebo

An inactive substance given to the control group in a research study.

pleural effusion (chylothorax)

Pleural effusion is a leakage and accumulation of fluid, chyle, into the chest cavity. Chyle is a milky-white bodily fluid circulated by the lymphatic system Chylothorax occurs when chyle leaks into the chest cavity surrounding the lungs.

pneumonitis

Inflammation of the lungs that can be a serious or rare complication of certain medications or infections.

pneumothorax

A collapsed lung characterized by pain and difficulty breathing due to air leaking into the space between the lung and the chest wall.

polycystic kidney disease (PKD)

A genetic disorder characterized by multiple cysts within each kidney. The cysts grow and multiply over time, leading to loss of kidney function. Because the TSC2 gene is adjacent to one of the PKD genes, a few individuals have both disorders, characterized by early-onset and severe PKD with various manifestations of TSC.

polymorphism

The occurrence of two or more different forms of a gene in the normal population.

positron emission tomography (PET)

PET scans detect nuclear radiation emitted by a "tracer" molecule ingested by or injected into a person to detect where the tracer molecule goes. The tracer used depends on what type of activity needs to be detected.

preclinical research

This refers to research performed on a drug before it can be tested in humans in a Phase 1 clinical trial. Preclinical research typically includes testing in animal models.

protein

Large biomolecules comprised of amino acids. Cells create a protein beginning with "instructions" encoded by a gene within the cell's DNA.

protoco

The written description of how a clinical study or experiment will be conducted, including the study's objectives, design and methods.

psychiatry

Psychiatry is the branch of medicine for physicians focused on the diagnosis, treatment and prevention of mental, emotional and behavioral disorders.

psychology

Psychology is the study of the mind and behavior. Psychologists are professionals but not necessarily physicians.

randomization; randomized

Method of distributing participants by chance to all arms of a clinical trial.

rapamycin

Rapamycin is a large molecule discovered in a soil sample from Easter Island in 1975. It is now manufactured and marketed as a drug named sirolimus and is approved by FDA to treat LAM.

rare disease

A rare or orphan disease is one that affects a small proportion of the population. In the United States, a rare disease is defined as a condition that affects fewer than 200,000 people. In the European Union, it is defined as a disease that affects fewer than 1 in 2,000 people. There are more than 6,000 rare diseases.

refractory epilepsy

See: intractable epilepsy

regulatory body

Governmental organization responsible for enforcing laws concerned with protecting the public's interest. Examples: Food and Drug Administration (FDA), European Medicines Agency (EMU).

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renal cell carcinoma

Also known as "kidney cancer" or "renal adenocarcinoma," renal cell carcinoma (RCC) is a malignant, often aggressive, tumor in the kidney. Fat-poor angiomyolipomas can appear similar to RCC on an MRI, so it is important to obtain an accurate diagnosis because the risks and treatments of each are very different.

respite care

Planned or emergency temporary care provided to caregivers of a person (child or adult) who is unable to care of themselves.

retinal hamartoma

A non-malignant tumor in the retina of the eye commonly associated with TSC.

Schedule I. II. III. IV. V

The DEA classifies drugs/substances into one of five schedules depending on the drug's medical use and its abuse or dependency potential. Schedule I substances are considered to be the most abuse-prone and least valuable medically. Schedule V drugs have relatively low potential for abuse. Most drugs have little abuse potential and are not scheduled.

sclerotic bone lesion

An abnormal increase in density, hardening or thickening of the bone.

seizure

Abnormal and unusually intense bursts of electrical activity in the brain, usually of sudden onset, that may cause strange sensations, emotions, behaviors, muscle spasms and possible loss of consciousness.

shagreen patch

Abnormal patches of skin resembling an orange peel, usually found on the lower back or on the back of the neck. Shagreen patches may be present on other parts of the body as well.

sirolimus (Rapamune®)

See rapamycin.

soft fibromas (skin tags)

Some individuals with TSC may have soft growths on the neck, trunk, armpits or groin. They may also have smooth, skin-colored bumps on the neck, armpits and near the flexure of the limbs.

specialty pharmacy

Specialty pharmacies manage, dispense and distribute high-cost, difficult-to-store, or highly regulated medications.

sporadic mutation

A sporadic mutation is a mutation in DNA not inherited from a person's parents. In TSC, two-thirds of individuals with the disease have no familial history of TSC. The other one-third of patients can trace their inheritance to an affected parent.

standard of care

Treatment that is accepted by medical experts as a proper treatment for a certain disease and that is widely used by healthcare professionals.

status epilepticus

A single epileptic seizure lasting more than five minutes or ≥2 seizures within a five-minute interval without the individual returning to his or her normal/baseline state between them. Can be of focal, generalized or unknown

subependymal giant cell astrocytoma (SEGA)

A nonmalignant tumor found in the brain of individuals with TSC. SEGAs typically grow near or in the ventricles and can cause hydrocephalus (increased pressure in the brain) if they block the flow of cerebrospinal fluid (CSF).

subependymal nodule

A non-cancerous collection of cells located along the edge of the brain's ventricles. Subependymal nodules can grow into SEGAs and/or become calcified (filled with a calcium deposit).

synapse

The area between two connected neurons. Neurotransmitters are typically exchanged in this area.

See TSC-associated neuropsychiatric disorders.

topical rapamycin

A mixture of rapamycin (sirolimus) in an ointment, cream, or gel which may be prescribed and applied directly to the skin to treat facial angiofibromas. A specific formulation of sirolimus topical gel (Hyftor™) was approved by the FDA in 2022. For many years, rapamycin has been compounded by pharmacists into various topical formulations.

transition age

The period during which a person shifts from childhood to young adulthood or from mid-late teens into the early 20s. This includes the age when an individual ceases to be the legal responsibility of a parent or guardian and assumes legally recognized adult rights and responsibilities, in addition to medical transitions from pediatric to adult specialists, educational transitions from high school to college or employment and other social, housing and employment transitions.

translational research

Translational research is the process of moving and applying knowledge gained from the basic sciences to its application in clinical and community settings, often summarized by the phrase "bench-to-bedside."

TSC-associated neuropsychiatric disorders (TAND); TAND Checklist

TAND includes aggressive behaviors, autism spectrum disorders, intellectual disabilities, psychiatric disorders neuropsychological problems, and school and occupational difficulties that are often expressed by individuals with TSC. A result of the 2012 TSC Clinical Consensus Conference was the creation of the TAND Checklist as a tool to routinely evaluate TSC's cognitive, behavioral and psychiatric impact on an individual.

TSCure

An international research collaboration focused on early intervention and prevention of TSC manifestations in children and adults.

TSC Biosample Repository

The TSC Biosample Repository is operated by the TSC Alliance to collect and distribute DNA from cheek cells, blood samples and tissue taken from surgeries.

tuberin

Tuberin is the protein encoded by TSC2. With the product of TSC1, hamartin, it forms the TSC protein complex, a group of proteins that, together, regulate mTORC1 activity.

ultrasound

A procedure that uses high-energy sound waves to look at tissues and organs inside the body. The sound waves make echoes that form pictures of the tissues and organs on a computer screen (sonogram).

ungual fibroma

Benign fibrous tumors found in the areas around the fingernails and toenails.

urology

Study of the male and female urinary tract system including kidneys, adrenal glands, ureter, bladder, urethra and the male reproductive organs.

vagus nerve stimulation (VNS)

An implanted medical device that involves delivering electrical impulses to the vagus nerve as an adjunctive treatment for certain types of intractable epilepsy.

variant of unknown significance (VUS)

A mutation or change in a gene sequence that is neither known to be disease-causing nor known to be found in people without a particular disease.

vigabatrin (Sabril®, Vigadrone®)

Antiepileptic drug that inhibits the breakdown of GABA. It is considered first-line therapy for infantile spasms associated with TSC. Generic versions of vigabatrin became available in 2017.

visual field

The entire area a person can see when his or her eyes are fixed in one position, including the peripheral (side) area.

visual field loss

When used in reference to vigabatrin, visual field loss refers to progressive loss of peripheral (side, top or bottom) vision, starting on the outside and working its way in.

white matter

Lighter colored tissue within the brain or spinal cord consisting primarily of neuronal axons and surrounding glial cells.

Wood's lamp

An ultraviolet light (UV, or blacklight) used to examine skin for hypopigmented macules in a darkened room.

zebrafish (Danio rerio)

A small freshwater fish commonly used in biological research as an animal model.

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Seizure Terms 101

(Adapted courtesy Epilepsy Foundation)

Naming Structure:

where seizures originate in the brain + awareness [if focal onset] + symptoms

focal onset: instead of partial; "focal" refers to where in the brain the seizure begins. Focal seizures can start in one area or group of cells in one side of the brain.

focal onset aware: individual is awake/aware during a seizure—formerly: simple partial seizure.

focal onset impaired awareness: individual is confused/awareness is affected in some way during a focal seizure—formerly: complex partial seizure

focal to bilateral tonic-clonic seizures: specific type of seizure that spreads from one side of the brain (focal) to both (bilateral) while phasing from tonic (rigid muscles) to clonic (muscles contract/relax rapidly) motor features—formerly: secondarily generalized seizure

generalized onset: all generalized seizures affect a person's awareness or consciousness in some way so no need to specify "impaired awareness"—generalized onset seizures affect both sides of the brain from the beginning of the event.

unknown onset: if a seizure's onset/beginning is not known or not observed (e.g., individual alone/asleep) it can be described as unknown (and later reclassified if appropriate)

Seizure Symptoms:

motor symptoms:

- clonic: rhythmic, jerking motions
- tonic: tense/rigid muscles
- tonic-clonic (formerly: grand mal)
- myoclonic: brief muscle twitching
- · myoclonic-tonic-clonic new term
- myoclonic-atonic
- · atonic: muscles that become weak or limp
- epileptic spasms: body flexes and extends repeatedly
- automatisms: repeated, automatic movements, e.g., clapping, rubbing hands together, lip smacking, chewing, etc.
- hyperkinetic: agitated thrashing or leg pedaling movements

nonmotor symptoms: (formerly: absence symptoms)

- typical absence: (formerly: petit mal)
- atypical absence: slow duration/termination of absence
- myoclonic absence
- eyelid myoclonia
- behavior arrest: cessation of motor activity

Quick Conversion:

Old Term	New Term
absence seizure	generalized absence
atonic / drop attack	focal/generalized atonic
complex partial	focal impaired awareness
simple partial	focal aware

Conference Acknowledgements

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Greg Grunberghosts The Care Giver

A series about navigating the complexities of living with rare forms of epilepsy

Greg Grunberg, an actor and advocate for his son living with a seizure disorder, is taking a trip across the country to connect with caregivers and hear their incredible stories.

Join Greg on an incredible journey of care

- Meet caregivers of families living with rare forms of epilepsy
- ✓ Hear stories from diagnosis to treatment
- ✓ Find out how other families have advocated for their loved ones
- Learn the importance of taking the time to care for yourself



These stories will bring you hope and strength in knowing you're not alone on this journey.



Scan the QR code to watch the series or visit

TheCareGiverSeries.com





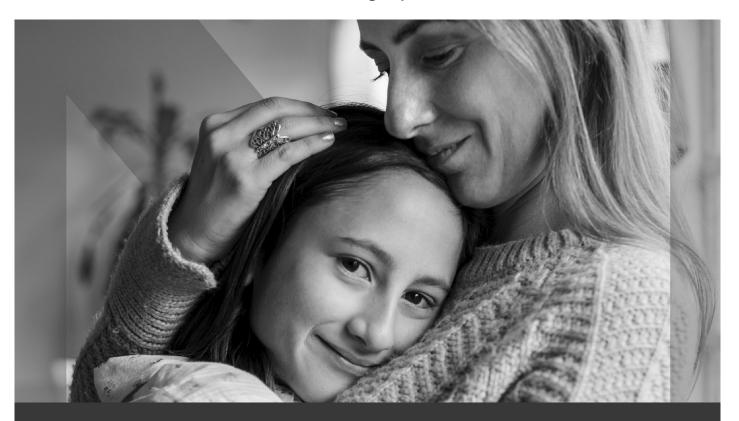
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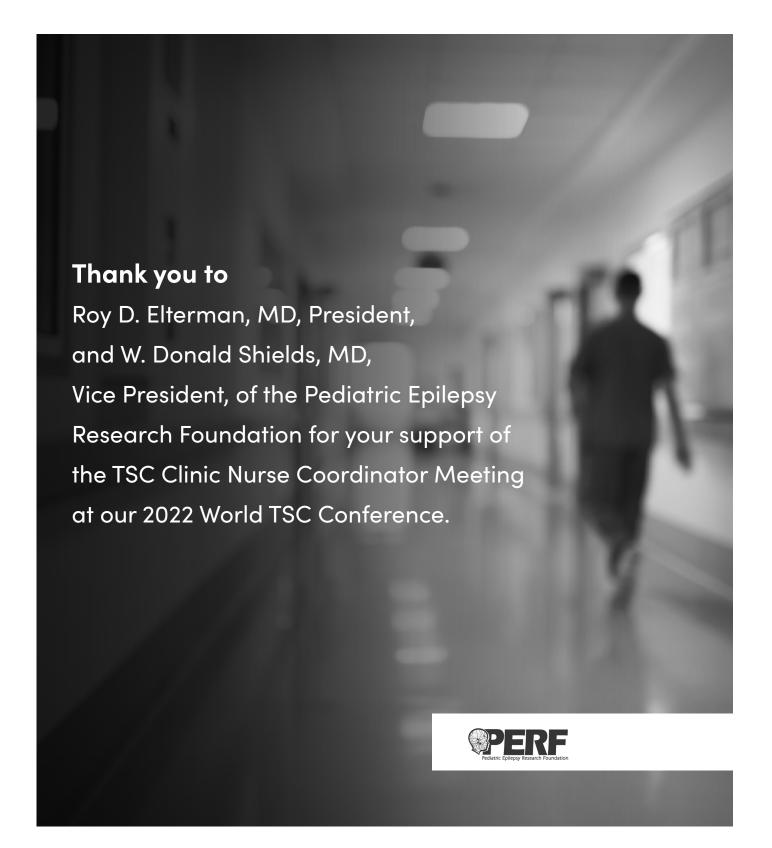
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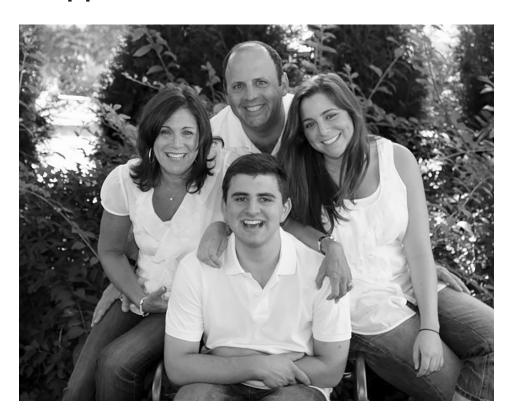
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The Bcureful Travel Fund at the TSC Alliance underwrites the TSC Travel and Lodging Patient Assistance Program, which is administered by the National Organization for Rare Disorders (NORD). This travel assistance program provides patients diagnosed with TSC with financial assistance for travel and lodging related to participation in a clinical trial and/or furtherance of treatment at a TSC Alliance-recognized Center of Excellence or TSC Clinic.

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The Parkes family has managed the challenges of TSC with determination and grace for more than 30 years!

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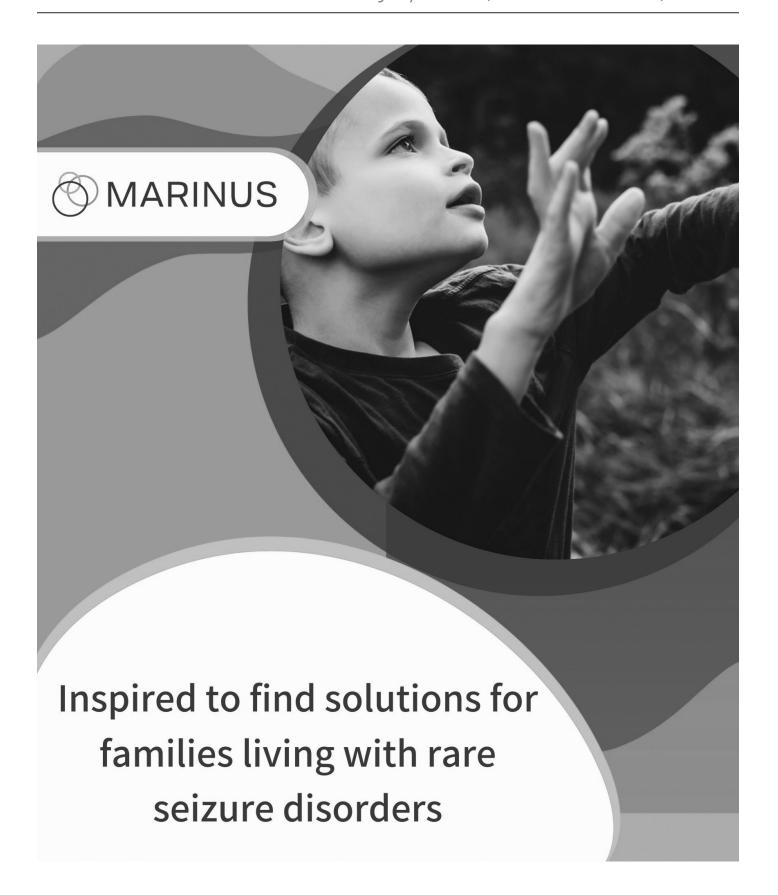
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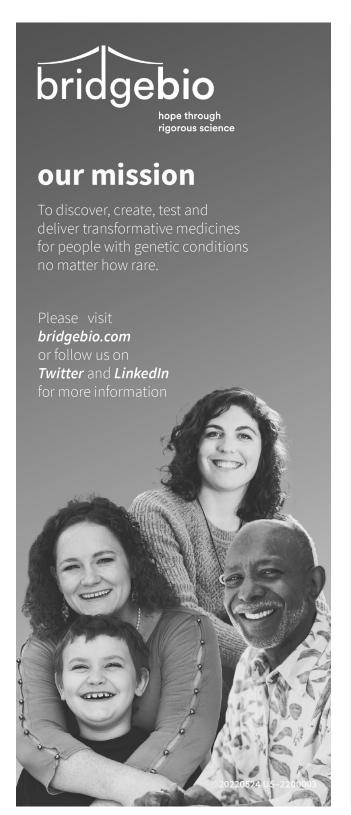


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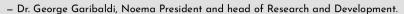








"Noema is committed to its partnership with the TSC community and has initiated the Galene Phase IIb study with Basimglurant (NOE-101), a candidate therapy aiming at controlling seizures in patients with TSC. Our lead molecule, Basimglurant, is a highly selective, potent and cell penetrant negative allosteric modulator of mGlu5 receptors. It has demonstrated a promising effect in animal models that represent the same features as the human condition and a good safety profile and is expected to transform the lives of patients living with TSC."







NOE-101

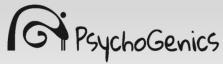
NOE-101 mGluR5 NAM (basimglurant)
For the management of seizures associated with tuberous sclerosis complex (TSC)
Clinical stage: Phase 2b



Ovid Therapeutics is honored to be part of this remarkable community. Our team is inspired by every family, and the work of the TSC Alliance and the community. Together we are strong. Thank you for including Ovid in this important event.

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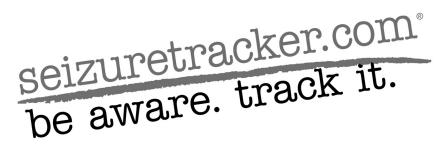


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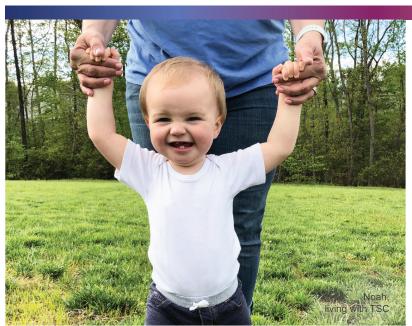
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