

# Consensus Guidelines: What You Need to Know

Chris Kingswood, MBBS, FRCP

Hope Northrup, MD

Darcy Krueger, MD, PhD

Pre-Conference Webinar

May 11, 2026



# Today's Moderator



**Chris Kingswood, MB, BS, FRCP**

# Housekeeping



- Cameras and microphones have been disabled for this webinar.
- Questions can be submitted via the Q+A button below throughout the presentation.
- This webinar is being recorded. The recording will be sent to all registrants and posted to YouTube and the TSC Alliance website.

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## Seizure Types in TSC

Wednesday, June 3, 2026, at 12:00 pm ET

## Genetics Made Simple

Wednesday, June 17, 2026, at 8:00 pm ET

## mTOR Inhibitors: Past, Present and Future

Tuesday, June 30, 2026, at 8:00 pm ET

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# Introduction

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# Why Guidelines?

- For the detailed instruction of doctors still learning
- For the guidance of those more expert

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# Why Guidelines?

- For the detailed instruction of doctors still learning
- For the guidance of those more expert
- Signal to payers what needs funding and to families what needs attention.
- Knowledge and treatment is always changing and improving

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# Today's presenters



**Hope Northrup, MD**



**Darcy Krueger MD, PhD**

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# What are Consensus Guidelines

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# Consensus Conference



- Definition (Cambridge Dictionary): a meeting at which experts from different specialist areas discuss a scientific or medical subject in which they are all involved, and try to reach agreement

# Background

- Original TSC Consensus Conference 1998 with 2<sup>nd</sup> TSC Consensus Conference 2012 (80 experts from 14 countries)
- All Chairs and Working Group Co-Chairs from 2012 Conference areas were invited to meet face-to-face at 2018 TSC World on Dallas, TX, July 25-26
- Pre-work included reviewing all new literature published since 2013 which was then discussed in working group meetings (telephone, email) prior to face-to-face meeting at TSC World 2018
- At face-to-face meeting, changes/updates/additions to Diagnostic Criteria and Surveillance and Management Guidelines determined
- Output published in *Pediatric Neurology* 123 (2021) pp 50-66  
**AVAILABLE** for those who want **ALL** details

# Diagnostic Criteria –Why Do We Need/Want?



- Rapid diagnosis and consistent characterization of disease
- Determine extent of phenotype
- Must be updated as new information is discovered

# TSC Consensus Guidelines scope

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# Diagnostic Criteria Updates

## 11 Major Features

- Hypomelanotic macules ( $\geq 3$ ; at least 5mm diameter)
- Angiofibroma ( $\geq 3$ ) or fibrous cephalic plaque
- Ungual fibromas ( $\geq 2$ )
- Shagreen patch
- Multiple retinal hamartomas
- ~~Cortical dysplasias ( $\geq 3$ )\*~~  
**(Multiple cortical tubers and/or radial migration lines)**
- Subependymal nodule ( $\geq 2$ )
- Subependymal giant cell astrocytoma
- Cardiac rhabdomyoma
- Lymphangiomyomatosis (LAM)\*\*
- Angiomyolipomas ( $\geq 2$ )\*\*

## 7 Minor Features

- “Confetti” skin lesions
- Dental enamel pits ( $\geq 3$ )
- Intraoral fibromas ( $\geq 2$ )
- Retinal achromic patch
- Multiple renal cysts
- Nonrenal hamartomas
- **Sclerotic bone lesions**

# Diagnostic Criteria: Genetic Testing



Recommendation: A pathogenic mutation\* in *TSC1* or *TSC2* is diagnostic for TSC

\*Most TSC-causing mutations are sequence variants that clearly prevent *TSC1* or *TSC2* protein production. Some mutations compatible with protein production (e.g. some missense changes) are well established as disease causing. Other variants should be considered with caution.

No changes to the recommendations from 2012

**HOWEVER**

Greater recognition of variability in TSC and more powerful testing options

# Updated Surveillance and Management Recommendations



- Provided for: newly diagnosed AND already diagnosed (following)
- One to two highlights per organ system
- Full article available on line free thanks to *Pediatric Neurology* (Dr. Steve Roach Editor)
- Map to other sessions on the organ system

# Updated Surveillance and Management Recommendations- Overall

- Increased emphasis on early screening for EEG abnormalities
- Enhanced surveillance and management of TSC-associated neuropsychiatric disorders
- New medication approvals

# Why Guidelines Matter

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# Clinical guidelines need to be actively used, not just written, to improve outcomes



# Why Use of Guidelines Matter

Engaged patients have improved adherence, safety, and satisfaction



Better adherence



Enhanced safety



Greater satisfaction



# Why Use of Guidelines Matter

Patients and clinicians who understand guidelines can:



Make informed decisions about care options



Recognize appropriate vs unnecessary treatments



Better manage complex and chronic conditions



# How can TSC patients, caregivers, and clinicians best utilize the guidelines?

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# Beyond the Guidelines: How We Can Improve Healthcare for People with TSC Around the World

(Stuart *et al.* *Pediatric Neurology* 2021. 123:77-84)

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Contents lists available at ScienceDirect

Pediatric Neurology

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## Original Article

### Beyond the Guidelines: How We Can Improve Healthcare for People With Tuberous Sclerosis Complex Around the World



Clare Stuart, MPH <sup>a</sup>, Carla Fladrowski <sup>b, c</sup>, Jennifer Flinn, EdD <sup>d</sup>, Berit Öberg <sup>e</sup>, Angela Peron, MD <sup>f, g, h</sup>, Micaela Rozenberg <sup>c, i</sup>, Catherine A. Smith <sup>j, \*</sup>

<sup>a</sup> Tuberous Sclerosis Australia, Sydney, New South Wales, Australia

<sup>b</sup> Associazione Sclerosi Tuberosa ASP, Rome, Italy

<sup>c</sup> European Tuberous Sclerosis Complex Association, Wiesbaden, Germany

<sup>d</sup> Tuberous Sclerosis Canada Sclérose Tubéreuse, Ontario, Canada

<sup>e</sup> TSC Sverige, Stockholm, Sweden

<sup>f</sup> Child Neuropsychiatry Unit – Epilepsy Center, San Paolo Hospital, Department of Health Sciences, Università degli Studi di Milano, Milan, Italy

<sup>g</sup> Human Pathology and Medical Genetics, ASST Santi Paolo e Carlo, San Paolo Hospital, Milan, Italy

<sup>h</sup> Division of Medical Genetics, Department of Pediatrics, University of Utah School of Medicine, Salt Lake City, Utah

<sup>i</sup> Associação de Esclerose Tuberosa em Portugal, Lisbon, Portugal

<sup>j</sup> TSC Alliance, Silver Spring, Maryland

# Beyond the Guidelines: How We Can Improve Healthcare for People with TSC Around the World

(Stuart *et al. Pediatric Neurology* 2021. 123:77-84)

- **People with TSC need health care that implements clinical guidelines**
  - Awareness and knowledge of the guidelines
  - Access to imaging and treatments
  - Access to treatments



# Beyond the Guidelines: How We Can Improve Healthcare for People with TSC Around the World

(Stuart *et al. Pediatric Neurology* 2021. 123:77-84)



- **People with TSC need health care that has access to TSC expertise**
  - Health care professionals have basic level of up-to-date and balanced information about TSC
  - Health care professionals know where and how to seek advice and reviews from TSC experts, *and be willing to do so*

# Beyond the Guidelines: How We Can Improve Healthcare for People with TSC Around the World

(Stuart *et al. Pediatric Neurology* 2021. 123:77-84)



- **People with TSC need health care that is coordinated and integrated**
  - Facilitate connections between local health professions and build networks
  - Encourage and advocate for the development of TSC multi-disciplinary clinics and centers
  - Evaluate clinical tools that can improve care coordination



# Examples: How can patients and caregivers utilize care guidelines?



Translate guidelines into patient-friendly language and formats (including digital accessibility and integration)



Use guidelines as question tools



Compare and track personal care within guidelines

Preventative screenings  
Medication adherence  
Treatment response and adjustments/alternatives



# Examples: How can clinicians make guidelines actionable for patients?



Assess guideline familiarity and use by patients



Create handouts and infocharts to share with patients at clinical visits and encounters



Incorporate guidelines into shared decision making with patients



Embed guidelines prompts into care delivery workflows

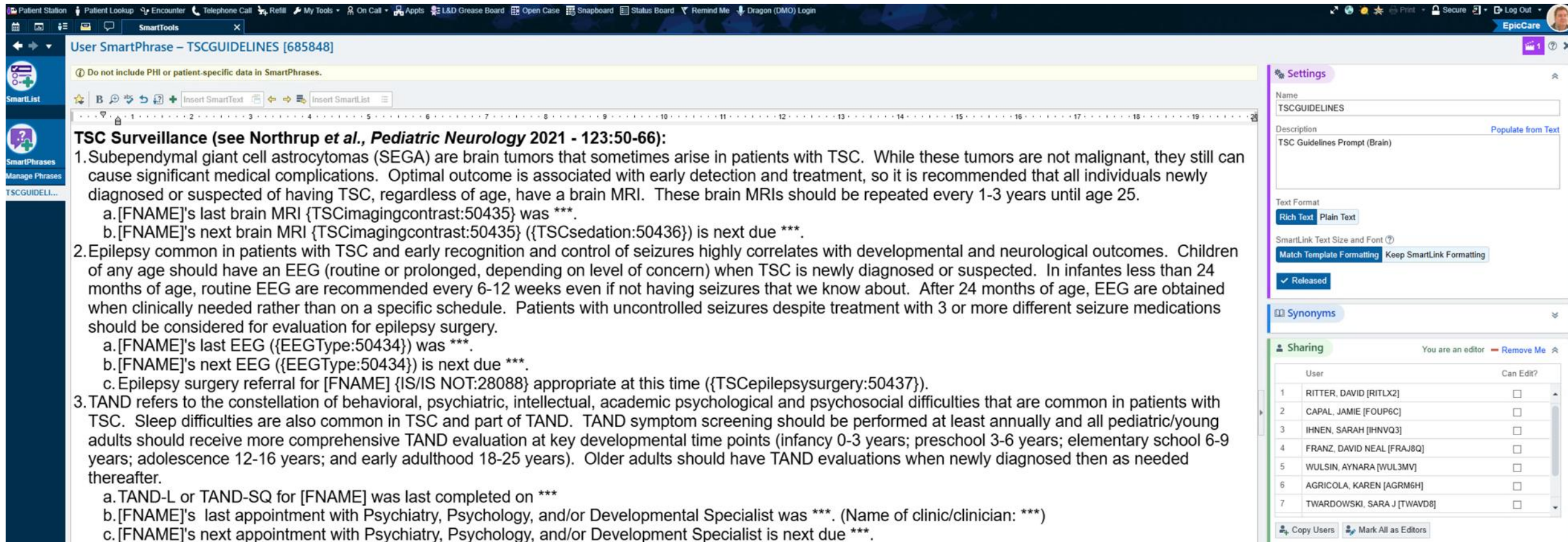
Electronic medical record smart phrases and flowcharts

Visit summaries and discharge instructions

# How can clinicians make guidelines actionable for patients



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**User SmartPhrase – TSCGUIDELINES [685848]**

Do not include PHI or patient-specific data in SmartPhrases.

**TSC Surveillance (see Northrup et al., *Pediatric Neurology* 2021 - 123:50-66):**

- Subependymal giant cell astrocytomas (SEGA) are brain tumors that sometimes arise in patients with TSC. While these tumors are not malignant, they still can cause significant medical complications. Optimal outcome is associated with early detection and treatment, so it is recommended that all individuals newly diagnosed or suspected of having TSC, regardless of age, have a brain MRI. These brain MRIs should be repeated every 1-3 years until age 25.
  - [FNAME]'s last brain MRI {TSCimagingcontrast:50435} was \*\*\*.
  - [FNAME]'s next brain MRI {TSCimagingcontrast:50435} ({TSCsedation:50436}) is next due \*\*\*.
- Epilepsy common in patients with TSC and early recognition and control of seizures highly correlates with developmental and neurological outcomes. Children of any age should have an EEG (routine or prolonged, depending on level of concern) when TSC is newly diagnosed or suspected. In infants less than 24 months of age, routine EEG are recommended every 6-12 weeks even if not having seizures that we know about. After 24 months of age, EEG are obtained when clinically needed rather than on a specific schedule. Patients with uncontrolled seizures despite treatment with 3 or more different seizure medications should be considered for evaluation for epilepsy surgery.
  - [FNAME]'s last EEG ({EEGType:50434}) was \*\*\*.
  - [FNAME]'s next EEG ({EEGType:50434}) is next due \*\*\*.
  - Epilepsy surgery referral for [FNAME] {IS/IS NOT:28088} appropriate at this time ({TSCepilepsysurgery:50437}).
- TAND refers to the constellation of behavioral, psychiatric, intellectual, academic psychological and psychosocial difficulties that are common in patients with TSC. Sleep difficulties are also common in TSC and part of TAND. TAND symptom screening should be performed at least annually and all pediatric/young adults should receive more comprehensive TAND evaluation at key developmental time points (infancy 0-3 years; preschool 3-6 years; elementary school 6-9 years; adolescence 12-16 years; and early adulthood 18-25 years). Older adults should have TAND evaluations when newly diagnosed then as needed thereafter.
  - TAND-L or TAND-SQ for [FNAME] was last completed on \*\*\*
  - [FNAME]'s last appointment with Psychiatry, Psychology, and/or Developmental Specialist was \*\*\*. (Name of clinic/clinician: \*\*\*)
  - [FNAME]'s next appointment with Psychiatry, Psychology, and/or Development Specialist is next due \*\*\*.

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# 2026 TSC Consensus Guideline Update Preview

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# 2026 International TSC Consensus Group Recommendations

- Broad international representation including clinicians, researchers, patients, and caregivers
- Comprehensive professional literature search
- Standardized evidence scoring (NCCN/GRADE)
- Standardized process for consensus formation (Delphi)
- Deliverables:
  - Summary consensus recommendations
  - Specialty-specific detailed manuscripts
  - Patient and clinician summaries and materials



# 2026 International TSC Consensus Group Recommendations



## Working Groups

Nephrology	Pulmonology	Genetics	Maternal and Fetal Medicine	Cardiology
Endocrinology	Gastroenterology	Ophthalmology	Dermatology	Dentistry And Oral Health
TAND	Epilepsy	SEGA	Radiology	Care Integration and Patient Perspectives

# 2026 International TSC Consensus Group Recommendations



2025

- September: Steering Committee organized, Working group leaders invitations
- October: Methods/Process refined, Working group leaders invited
- November: Methods/Process refined, Working group leaders invited
- December: Launch meeting with Working group leaders

2026

- January: Working group participants invited, Delphi Survey #1, Literature review
- February: Working group meetings
- March: Working group meetings
- April: Working group meetings
- **May: Working group meetings**
- June: Working group meetings
- July: World Conference deliberations, Recommendations #1, Delphi Survey #2
- August: Working group meetings, Recommendations #2, Delphi Survey #3
- September: Draft final recommendations manuscript
- October: Publish final recommendations manuscript
- November: Draft specialty-specific manuscripts
- December: Publish final specialty-specific manuscripts

# Q & A

Moderated by Chris Kingswood, MBBS, FRCP

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# Genetics



## Pre-Conference Webinar: Genetics Made Simple

- Wednesday June 17 from 8-9:00PM ET on Zoom
- *Moderator:* Danielle Nolan, MD
- *Speaker:* Hope Northrup, MD

## Reproductive Decision Making and Childbearing Considerations

- Friday July 31 from 10:30 AM-Noon in Maple 3
- *Moderator:* Heather Radtke, MS, CGC
- *Speakers:* Iris Mustich, MPH; Lara Friel, MD, PhD; Jennifer Glass, LGC; Lyndsay Harshman, MD, MS

# Genetics, cont'd



## Future Role of Genetics in Personalized Medicine

- Saturday, August 1 from 10:30AM-12PM in Adams A
- *Moderator:* Hope Northrup, MD
- *Speakers:* Angela Peron, MD, PhD; Laura Farach, MD; Jeffrey Calhoun, PhD

## Genetic Therapies

- Saturday August 1 from 1-2:30PM in Adams A
- *Moderator:* Elizabeth Thiele, MD, PhD
- *Speaker:* Julian Martinez-Agosto, MD, PhD; Clayton Beard, PhD; Mark Hester, PhD

# Women's Health



## Fetal and Perinatal Health

- Friday, July 31 from 1PM-2:30PM in Maple 4
- *Moderator:* Heather Radtke, MS, CGC
- *Speakers:* David Ritter, MD, PhD; Sarah Weatherspoon, MD; Stephanie Randle, MD, MS, Allison Divanovic, MD

## Health Issues for Women with TSC

- Saturday August 1 from 1-2:30PM in Maple 5
- *Moderator:* Lisa Henske, MD
- *Speaker:* Lara Friel, MD, PhD; Mark Keezer, MDCM, PhD; Kate Richardson, MS, CGC

# Brain and Tumors



## Preparing Your Family for Epilepsy Surgical Evaluation and Surgery

- Friday July 31 from 1-2:30PM in Spruce 3
- *Moderator:* Brenda Porter, MD, PhD
- *Speakers:* Lesley Holmes, JD; Emilie Hyde; S. Katie Ihnen, MD, PhD; Monika Jones, JD

## Epilepsy Surgery and Brain Imaging

- Friday July 31 from 3-4:30PM in Adams A
- *Moderator:* Stephanie Randle, MD, MS
- *Speakers:* Jeffrey Blount, MD, MPH; Aria Fallah, MD, MS; Jonathan Roth, MD; Wenbo Zhang, MD

# Brain and Seizures



## Pre-Conference Webinar: Seizure Types in TSC

- Wednesday June 3 from 12-1:00PM on Zoom
- *Moderator:* Shelly Meitzler
- *Speakers:* Sam Amin, MBCHB, MRCPCH, MSc, PhD; E. Martina Bebin, MD, MPA;

## Seizure Treatments

- Thursday July 30 from 2:45-4:15PM in Adams A
- *Moderator:* Priya Tatachar, MD
- *Speakers:* Sam Amin, MBCHB, MRCPCH, MSc, PhD; Peter Crino, MD, PhD; Thomas Foutz, MD, PhD; Rod Foroozan, MD; Darcy Krueger, MD, PhD

# Brain and Seizures, cont'd



## Seizure Action Plans and Rescue Medications

- Friday July 31 from 1-2:30PM in Spruce 3-4
- *Moderator:* Vijay Vishwanath, MD, PhD
- *Speakers:* Rob Moss; Raj Rajaraman, MD, MS; Irfan Sheikh, MD

## SUDEP Risk and Seizure Monitoring Technology

- Saturday August 1 from 1-2:30PM in Maple 3
- *Moderator:* James Wheless, MD
- *Speakers:* Peter Crino, MD, PhD; Mary Duffy; Bernice Martin Lee; Donald Phillips, MD, MPH, FACNS

## Refractory Seizure Management and Tools

- Saturday August 1 from 3-4:00PM in Maple 4
- *Moderator:* Rohini Coorg, MD
- *Speakers:* Sam Amin, MBCHB, MRCPCH, MSc, PhD; Irfan Sheikh, MD; Rob Moss; James Wheless, MD

# TAND



## TSC Community-Driven Research and Global TAND Collaborations

- Friday, July 31, 8:30-10AM in Adams A
- *Moderator:* Steven L. Roberds, PhD
- *Speakers:* Dean Aguiar, PhD; Anita Bhatia, MHA; Liz Cassidy, MPH; Petrus de Vries, MBChB, FRCPsych, PhD; Agnies van Eeghen, MD, PhD

## Dysregulated Behaviors

- Friday, July 31, 10:30AM-12PM in Adams A
- *Moderator:* Tanjala Gipson, MD
- *Speakers:* Jamie Capal, MD, Peter Davis, MD, Lauren Shillinger, Sean Shillinger

## Organizing Your Thoughts and Emotions and Managing Relationships (only open to adults with TSC)

- Friday, July 31, 10:30AM-12PM in Spruce 2
- *Moderators:* Shannon and Rob Grandia
- *Speakers:* Danielle Clark, MPH, RS, RD, LD; Petrus de Vries, MBChB, FRCPsych, PhD

# TAND, cont'd

## Pre-Puberty and TAND

- Friday, July 31, 1-2:30PM in Maple 5
- *Moderator:* Mustafa Sahin, MD, PhD
- *Speakers:* Peter Davis, MD; Shafali Jeste, MD; Joanna Lomas-Mevers, PhD, BCBA-D

## Mood, Anxiety, and Overactive/Impulsive Behaviors

- Friday, July 31, 1-2:30PM in Maple 3
- *Moderator:* Anna Jansen, MD, PhD
- *Speakers:* Jamie Capal, MD; Alice Bryn Salter, MD; Agnies van Eeghen, MD, PhD

## Autism: Diagnosis, Therapy, and Managing Behaviors

- Friday, July 31, 3-4:30PM in Maple 3
- *Moderator:* Peter Davis, MD
- *Speakers:* Petrus de Vries, MBChB, FRCPsych, PhD; Shafali Jeste, MD; Nicole McDonald, PhD; Mustafa Sahin, MD, PhD



# TAND, cont'd

## Navigating Issues Living with TSC as an Independent Adult (TAND, Epilepsy, Surveillance)

- Friday, July 31, 3-4:30PM in Spruce 2
- *Moderator:* Derek Bauer, MD
- *Speakers:* Jamie Capal, MD; David Ritter, MD, PhD

## Behavioral Plans

- Saturday, August 1, 10:30AM-12PM in Spruce 3-4
- *Moderator:* Kim Hindery
- *Speakers:* Peter Crino, MD, PhD; Billy Holcombe, PhD; Joanna Lomas-Mevers, PhD, BCBA-D

## Overview of TAND Medications for Harmful and Disruptive Behaviors

- Saturday, August 1, 3-4:30PM in Maple 3
- *Moderator:* Mustafa Sahin, MD, PhD
- *Speaker:* Jay Salpekar, MD, FANPA

# Kidney



## Kidney Tumors, Cysts and Treatment Options

- Friday July 31 from 1-2:30PM in Adams A
- *Moderator:* Chris Kingswood, MD
- *Speakers:* John Bissler, MD; Lyndsay Harshman, MD, MS; Elahna Paul, MD, PhD; Joshua Samuels, MD, PhD;

## Managing Progressive Kidney Disease

- Friday July 31 from 3-4:30PM in Maple 4
- *Moderator:* Oded Volovelsky, MD, PhD
- *Speakers:* Elahna Paul, MD, PhD; Joshua Samuels, MD, MPH

# Lungs

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## Treatment Options and Clinical Trials in LAM

- Thursday July 30 from 2:45-4:15PM in Maple 4
- *Moderator:* Joel Moss, MD, PhD
- *Speakers:* Greg Downey, MD, FRCPC; Elizabeth (Lisa) Henske, MD; Simon Johnson, DM, FCRP; Gwen Pryor

## Living with LAM

- Friday July 31 from 3-4:30PM in Maple 5
- *Moderator:* Elizabeth (Lisa) Henske, MD
- *Speakers:* Cori Fratelli, MSN, FNP-C, RN; Nishant Gupta, MD; Joel Moss, MD, PhD

# Skin and Teeth

## Skin and Dental Manifestations and Treatment Options

- Saturday August 1 from 3-4:30PM in Maple 5
- *Moderator:* Thomas Darling, MD, PhD
- *Speakers:* Elizabeth Gosnell, DMD, MS; Deeti Pithadia, MD

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# Other – Liver, Pancreas, Heart, and Eyes

## Other Organ Involvement Through the Lifespan: Heart, Eyes, Liver and Pancreas

- Saturday August 1 from 10:30-12PM in Maple 4
- *Moderator:* Kate Richardson, MS, CGC
- *Speakers:* Rod Foroozan, MD; David Ritter, MD, PhD; David Rodriguez-Buritica, MD



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