

# Navigating the transition years of TSC

An information guide to navigate tuberous sclerosis complex (TSC) from 14 to 26 years of age.



## Foreword

These navigation guides were developed by the TSC Alliance® to provide essential information to help guide individuals and families through the complexities of tuberous sclerosis complex (TSC) across the lifespan. TSC uniquely affects each individual differently, therefore these guides complement the TSC Navigator tool ([www.tscalliance.org/tscnavigator](http://www.tscalliance.org/tscnavigator)) and focus on providing you with proactive material to help navigate the medical and non-medical aspects of this journey specific to the defined age group. While each developmental phase brings its own share of excitement and challenges, our guides are to complement your TSC journey and to empower you to live your fullest life.

### Navigation guides across the lifespan

- Navigating the early years of TSC (in utero to 5 years of age)
- Navigating the school-age years of TSC (6 years to 13 years of age)
- Navigating the transition years of TSC (14 years to 26 years of age)
- Navigating the adult years of TSC (27 years or older)

PDF versions of each guide with clickable links are available at [www.tscalliance.org/navigationguides](http://www.tscalliance.org/navigationguides).

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## Section One: My child was just diagnosed, now what?

A diagnosis of TSC can come at any point in life. With recent advancements and knowledge about TSC the diagnosis is coming earlier with onset of the more serious manifestations. If you are continuing from the previous guide and your loved one already has a TSC diagnosis, please feel free to skim through this section.

### What is tuberous sclerosis complex?

Tuberous sclerosis complex (TSC) is a genetic disorder that causes tumors to form in many different organs, primarily in the brain, eyes, heart, kidney, skin and lungs. TSC is a complicated disease. Some people live with few symptoms while others need continual support. Many people with TSC live independent, healthy lives and enjoy challenging professions such as medicine, law, education and research. The TSC journey is unique to each individual and family – even siblings and identical twins can have different experiences. Understanding the disease will give you hope, strength, and empowerment through all life stages.

Too often, TSC goes undiagnosed. But we know at least two babies born each day in the United States will have it. Nearly one million people worldwide are estimated to be living with TSC, with approximately 50,000 in the United States.

Most people with TSC will live a normal life span. Although there is no cure, there is hope. Research has proven that early diagnosis and intervention(s) are key for optimizing long-term outcomes. Advancements in research continue to deliver new and improved therapeutic options.

### Where do I go?

It is common for medical providers to be unfamiliar with TSC, including neurologists and pediatricians. It is crucial to have a local neurologist and pediatrician involved in your child's healthcare team. Finding a good fit is essential as these care team members will be your child's expert and, along with you, be your biggest advocate as you start your TSC journey.

During this age group, a pediatric neurologist who is familiar with TSC should be part of your child's healthcare team. Epilepsy (seizures) is frequently the main manifestation of TSC that brings upon the diagnosis, but not always. Even if your child presented with a TSC diagnosis and has not shown any signs of seizures, it is still clinically recommended to be evaluated during this phase as signs and symptoms of TSC change rapidly during these years due to the normal growth and development. As a parent you want nothing less than the best and connecting with a local or out-of-state TSC clinic to help guide clinical treatment options is appropriate. If you do not have access to a local TSC Clinic, you can also make an appointment with a pediatric neurologist experienced in epilepsy. Many TSC experts can offer provider-to-provider recommendations and our TSC Support Navigators can assist with this process.

Insurance coverage can also be a factor in ensuring your child does not have delayed intervention and care. It is important to consider if out-of-state healthcare is right for your family. For those who are considering traveling across state lines, please go to the insurance barriers page in the medical challenge section to learn more on how to proactively prevent issues from occurring. You can locate a TSC clinic by visiting the TSC Alliance website.

### Clinical management and your healthcare team

Being a successful caregiver doesn't mean you need to have all the answers or become an expert in TSC. You can empower yourself by clearly advocating your understanding of TSC and what you need to better understand, risk and benefit of treatment, short- and long-term outlook and how to prepare and balancing insurer- or pharmacy-based requirements.

Unfortunately, not every situation is ideal, and stress can lead to strong emotions. During those times it is important to focus on the goal you need to accomplish. Be realistic given the situation and options available to help redirect and find a solution to the current problem. Always remain calm when situations have escalated to anger and frustration. Most institutions and clinics have patient advocates. If you do not feel comfortable or agree with a treatment plan, you can always request to speak to an advocate prior to leaving (this is even true if you are in the emergency department!).

Because TSC can affect many different organ systems, you might find your child under the care of various medical specialists. Most of these specialties are separated between pediatric and adult patients. Two medical specialties are crucial to helping you manage and coordinate all these specialists: your pediatrician (children) or internist (adults). These clinicians are specially trained to manage the overall health of an individual, including helping you connect with and get the most out of your relationships with specialists.

Please reference the specialists you may encounter in this journey in the appendix of this section for more insight. Since TSC is a rare disease, sometimes you will have to travel out of state to see a TSC expert for clinical care or for those interested in clinical trials. The Bcureful Travel Fund is a travel assistance program that provides patients diagnosed with TSC with financial assistance for travel and lodging at TSC Alliance recognized centers. Please see the Bcureful Travel Fund flyer in the appendix of this section or online in the TSC Navigator.

## **Knowing your rights as the parent (how to advocate for your child in the medical setting)**

When caring for a medically complex child it's important you feel part of the decision-making process. You should be asking as many questions as it takes for you to feel comfortable and satisfied with the treatment plan. Here are some frequent questions you may want to ask your healthcare team:

- Do you have experience treating people with TSC or willing to work with a TSC expert?
- Are you aware of the 2021 consensus guidelines for treating TSC?
- What is the treatment plan you suggest for specific issue/manifestation?
- What tests should be done next and how frequently?
- How long should we wait to see if this medication/therapy works before we try a new one?
- Is this the only option available?
- What do we need to look for or be aware of during appointments?
- When should we visit you again?
- How can I contact you if above occurs? What are the afterhours / weekend process to contact you??
- What additional information can I provide that will help you offer the best care?
- Who is the best person for me to contact (name and phone number or email address) if I'm having issues with obtaining the medications you prescribed?

It is essential you find a provider who connects with your child and you as the caregiver. Having a medical professional who is open to conversation and answering questions is vital to a healthy medical journey. Your child's medical team should be helping you understand the treatment plan and any concerns at every appointment. Even in the toughest of situations it's important to stay respectful. Sometimes your provider will exhaust all resources and recommend a second opinion or additional insight from another expert. This does not mean you have to choose one or the other but offers additional input to ensure all options are being considered.

In some situations, families have found themselves not feeling heard or an equal part of the decision-making process. If you find yourself in this situation, it is okay to seek a second opinion. Most clinicians encourage second opinions, but do not always bring it up, so do not be afraid to have the conversation. Your current specialist needs to send medical records over to the new specialist or can help expedite a second opinion request so it's best to have him or her involved. In the unlikely scenario you find yourself feeling your child's care is at jeopardy for requesting a second opinion, then that is red flag on that providers practice and is not acceptable in clinical care.

Make sure you educate yourself as much as possible about TSC, you can find up-to-date information on the TSC Navigator and TSC Academy. Along with educating yourself about TSC, networking with other people in the TSC community is extremely valuable.

## Staying organized

Making appointments with multiple providers can be exhausting and it can be tough to predict how to prepare for new consultations to ensure the appointment is successful. Learn more on [How to Have a Successful Appointment and Prescription Management](#) on the TSC Navigator under [Coordination of Care](#); you can also find a copy in the appendix of this section.

Because this is the first phase of your journey, many laboratory and diagnostic tests will be mentioned along the way. Having an understanding of each will help you feel more empowered and confident as you have open conversations with your healthcare team. Knowing what to expect and when to proactively assess a situation is critical with this diagnosis. Building a solid foundation now will make a huge difference during your journey. Learn more on [Understanding Medication and Testing](#), which can be found in the appendix of this section.

Trying to stay organized when you are a parent or a caregiver to an individual requiring various appointments can be overwhelming. It is essential to be organized. Creating a medical binder, or notebook, where you can record medical information such as appointments and schedules will help you stay organized. In the appendix of this section, you will find sample logs and charts that have been helpful to others in the past. You can find downloadable versions in the TSC Navigator's [Resources](#) section.

## How do I get connected?

Along your journey, it is helpful to know you are not alone. Despite this being a very difficult time, the TSC community has compiled helpful resources to guide you during this phase. These tips and guides may not answer all your questions and our TSC Support Navigators are available to help you at any time.

This journey can feel lonely at times but connecting with another family that has navigated these steps before you can make all the difference. To gain more insight on caregiver selfcare please go to [caregiver mental health](#) under the resource section. In addition, there are plenty of ways you can connect with the TSC Community or reach out to our Support Navigators for assistance. Finding your local Community Alliance is an excellent place to start, please visit the TSC Navigator for more about a Community Alliance near you. For up-to-date information about the so TSC Alliance initiatives, videos, webinars, podcasts, information sessions, research, events, and community spotlight, you can find the TSC Alliance on social media platforms. If you feel like volunteering, there are also plenty of opportunities to align with any specific interest you feel is best suited for you.

### TSC Alliance social media channels: @tscalliance

- **Twitter:** [twitter.com/tscalliance](https://twitter.com/tscalliance)
- **Facebook:** [facebook.com/tscalliance](https://facebook.com/tscalliance)
- **Instagram:** [instagram.com/tscalliance](https://instagram.com/tscalliance)
- **YouTube:** [youtube.com/tscalliance](https://youtube.com/tscalliance)

### Facebook options

Connect with other members of the community in the private TSC Alliance Tuberos Sclerosis Complex Discussion Group on Facebook. This group offers support from members who are in all different stages of the TSC journey. In addition, Community Alliance Facebook pages, which are state- and/or region-specific, will allow you to connect to others in your area and learn about local events and gatherings.

Facebook also has other private support pages for TSC parents. To join these private groups, you can search for TSC Mommies ONLY – Tuberos Sclerosis Complex or TSC Daddies and request to join.

## Volunteer opportunities

Volunteering offers an opportunity to connect and engage with other families in the TSC community. There are various ways to participate as a volunteer with a position to fit anyone who is interested. For more information about a volunteer position please go to the TSC Alliance website for more information.

## Section One Appendix

### TSC Alliance resources

The TSC Alliance offers several digital resources to share the most up-to-date information on tuberous sclerosis complex (TSC) and educate our constituents about the disease, standards of care and ongoing research efforts. See a summary of these resources below.

| Resource                   | Description   | Website  |
|----------------------------|---|--|
| <b>TSC Navigator</b>       | <p>An easy-to-use, interactive online tool to help guide individuals and families through the complexities of tuberous sclerosis complex (TSC) across the lifespan, proactively manage their care and live their fullest lives. The goal of the TSC Navigator is to ensure families, caregivers and supporters of those impacted by TSC can find the right information at the right time.</p> <p><i>Please note: The TSC Navigator is a living resource, and some sections may be added, updated or changed as new or updated resources become available.</i></p> | <a href="http://www.tscalliance.org/tscnavigator">www.tscalliance.org/tscnavigator</a>   |
| <b>TSC Now</b>             | <p>A podcast from the TSC Alliance featuring conversations with leading TSC researchers, clinicians, families and individuals affected by TSC and others. Topics include the latest breakthroughs in TSC research, common challenges faced by those affected, upcoming TSC Alliance initiatives, TSC treatment options and living with TSC. Listen and subscribe wherever you listen to podcasts.</p>   | <a href="http://tsc-now.blubrry.net">tsc-now.blubrry.net</a>   |
| <b>TSC Matters</b>         | <p>A bimonthly community e-newsletter from the TSC Alliance highlighting TSC research updates, TSC clinical trials, upcoming TSC Alliance events and more.</p>  | <a href="http://www.tscalliance.org/tsc-matters">www.tscalliance.org/tsc-matters</a>   |
| <b>TSC Academy</b>         | <p>An online educational platform created for the TSC community. The platform features lessons about the basic, preclinical and clinical science of TSC; how research helps us understand and treat the disease; how the TSC Alliance, Federal, state and local governments and other stakeholders work together to find a cure; and how you can play a role.</p>   | <a href="http://www.tscacademy.org">www.tscacademy.org</a>   |
| <b>Videos and Webinars</b> | <p>The TSC Alliance hosts regular educational webinars on a variety of TSC related topics of interest featuring known experts in the TSC community. These webinars include live question-and-answer opportunities with these experts.</p>   | <a href="http://www.tscalliance.org/individuals-families/webinars-and-videos">www.tscalliance.org/individuals-families/webinars-and-videos</a> |

**Other notable resources**

| <b>Organization</b>                                | <b>Description</b>   | <b>Website</b>   |
|--|--|--|
| <b>Center for Parent Information and Resources</b> | Family friendly information and research-based materials on key topics for Parent Centers. Supporting the Parent Centers who serve families of children with disabilities.   | <a href="http://www.parentcenterhub.org">www.parentcenterhub.org</a>                   |
| <b>The Arc</b>                                     | Promotes and protects the human rights of people with intellectual and developmental disabilities and actively supports their full inclusion and participation in the community throughout their lifetimes.  | <a href="http://www.thearc.org">www.thearc.org</a>                                     |
| <b>Patient Advocate Foundation (PAF)</b>           | Provides professional case management services to patients with chronic, life threatening and debilitating diseases.   | <a href="http://www.patientadvocate.org">www.patientadvocate.org</a>                   |
| <b>Family Voices</b>                               | Is a national organization and grassroots network of families and friends of children and youth with special health care needs disabilities that promotes partnership with and families — including those of cultural, linguistic, and geographic diversity— to improve healthcare services and policies for children. | <a href="http://www.familyvoices.org">www.familyvoices.org</a>                         |
| <b>Epilepsy Foundation</b>                         | Connects the people, data and resources needed to address challenging health problems associated with seizures and epilepsies — and promotes education, policy, research and systemic change that will improve the life of those living with epilepsy.   | <a href="http://www.epilepsy.com">www.epilepsy.com</a>                                 |
| <b>Child Neurology Foundation</b>                  | Serves as a collaborative center of education, resources, and support for children and their families living with neurologic conditions and facilitates connection with medical professionals who care for them  | <a href="http://www.childneurologyfoundation.org">www.childneurologyfoundation.org</a> |
| <b>PACER Center, Inc.</b>                          | Enhances the quality of life and expands opportunities for children, youth and young adults with all disabilities and their families so each person can reach his or her highest potential.  | <a href="http://www.pacer.org">www.pacer.org</a>                                       |



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## Medical Specialists

Because TSC can affect many different organ systems, you might find yourself working with many different medical specialists. Most of these specialties are separated between pediatric and adult patients. There are two medical specialties that are crucial to helping you manage and coordinate all of these specialists: your **pediatrician** (children) or **internist** (adults). These clinicians are specially trained to manage the overall health of an individual, including helping you connect with and get the most out of your relationships with specialists.

### Cardiologist

Specializes in the diagnosis and treatment of conditions involving the heart.

### Dermatologist

Specializes in the diagnosis and treatment of conditions involving the skin – some, but not all, are trained to treat the skin manifestations of TSC.

### Epileptologist

Specializes in the diagnosis and treatment of individuals who have epilepsy – these are physicians who completed an additional round of training in epilepsy following completion of their neurology training.

### Geneticist

Specializes in the diagnosis of genetic conditions and provides recommendations about follow up care.

### Genetic Counselor

Trained to assist individuals who require genetic testing as well as providing guidance for reproductive decision making – typically not MDs or DOs, but specialists with a master's degree (or doctorate) in genetic counseling and a professional license.

### Gynecologist

Specializes in the diagnosis and medical/surgical treatment of diseases involving female reproductive organs. Also provides overall care and routine management of female reproductive organs.

### Nephrologist

Specializes in the diagnosis and treatment of diseases involving the kidney.

### Neurologist

Specializes in the diagnosis and treatment of disorders relating to the central nervous system (brain, spinal cord, and all the nerves throughout the body).

### Neurosurgeon

Specializes in surgical procedures that involve the central nervous system. Often, they will further subspecialize and only perform certain types of surgeries (i.e., epilepsy surgery, brain tumor resection, etc.).

### Neuropsychiatrist

Specializes in the management of mental health symptoms that are attributable to diseases of the central nervous system.

### Oncologist

Specializes in the diagnosis and treatment of cancer.

### Ophthalmologist

Specializes in the diagnosis and medical/surgical treatment of eye disorders.

### Psychiatrist

Specializes in the diagnosis and management of mental health disorders.

### Pulmonologist

Specializes in the diagnosis and treatment of diseases that involve the lungs.

### Urologist

Specializes in the diagnosis and medical/surgical treatment of diseases involving the bladder and lower urinary tract of both sexes. Also provides overall care and routine management of the prostate and male reproductive organs.



## Checklist for Successful Appointments and Prescription Management

Not quite sure what to ask or say during your next appointment? Here are some prompts and tips to help you prepare.

- What are my top concerns to address at this appointment?  
**TIP:** Write down questions and topics to discuss on a slip of paper or on your smartphone notes app so you don't forget them.
- Do I have any videos to show of seizures or any other strange behaviors/occurrences?  
**TIP:** Queue any videos up before your appointment so you don't have to spend time scrolling through your phone to find them. Consider creating an album or folder on your phone to keep important videos.
- Find out who you should contact at your physician's office if you have any follow up questions or concerns
- Make sure to jot down any key steps for **you** to take following this appointment
  - Next scan (MRI, EEG, CT, etc.):
  - Next lab work:
  - Are there any forms you need to fill out before scheduling tests?
  - Do I need any tests or bloodwork done to monitor any of the medications I am on?
- Make note of any test results that are shared with you at the appointment and make sure you're able to access them via a secure web portal or by asking for a paper copy
- Make a plan for your next appointment – when and how to schedule it.
- What medications does this doctor prescribe?
  - How many refills do I have left?
  - Will these refills last until the next appointment?
- Do any of these medications require a prior authorization (PA)?
  - How long would the PA be approved for?  
**TIP:** Set a reminder on your phone to go off at least two weeks before the prescription expires to remind you to start the PA process
- Make a note of the following important information for your prescription:
  - Pharmacy Name:
  - Address:
  - Phone & Fax Numbers:
  - Point of contact at your provider's office in case anything goes awry with filling your prescription:  
**TIP:** Save the pharmacy as a contact in your phone for easy access later.
- Determine what the estimated turnaround time is for prescription requests – especially if it's different for submissions via a secure web portal vs. requests by phone.  
**TIP:** Request as soon as you can to refill a prescription (retail or specialty) – if it's too soon to refill, ask the pharmacist when is the soonest you can make a request. Also, pay attention to weekends and holidays and try to avoid needing a refill around those times, if possible.



## Medication and Testing

Some treatment plans for TSC may be daunting. Here are some prompts to help facilitate a dialogue with your provider to help you feel confident about their intended treatment and surveillance plan.

### Initial questions to ask

- What is this test or medication for?
- Why is this test or treatment a good option for me, and what risks are there to consider?
  - Are there any potential complications I should consider?
  - What are the side effects of this medication?
    - If there are potential side effects, what are my options for managing them?
    - Who do I call, or where do I go, if I begin noticing or experiencing concerning symptoms?
- How long will it take for the medication to start working?
  - What does a "titration phase" mean?
  - What is a "loading dose" and when do we plan on dose-reducing?
  - If this is an anti-seizure medicine, what's an acceptable number of breakthrough seizures – when should I call?
  - What do I need to do if I miss a dose of this medication?
- Are there any other treatment options?
  - If not, what are the risks and benefits of waiting?
  - If I elect to decline this test or medication, what signs or symptoms should I be aware of to reconsider?
- Do you have any recommendations for a second opinion?
  - **SIDENOTE:** it is a myth that providers get upset when asked about second opinions. Second opinions can be a crucial affirming step when you are faced with an important decision, and many healthcare providers appreciate input from their peers, a second set of eyes.

### Testing and results

- What would be achieved by knowing the results of this test?
- Can you help me understand the accuracy of this test?
- Will there be another follow-up test, depending on findings from the initial?
- When will I receive test results? Who will discuss them with me?

### Insurance

- Will my insurance cover this test or treatment?

### Lifestyle changes

- Are there any other considerations I should be aware of before starting this medication? Should I plan to...:
  - Change my diet?
  - Notice any changes to sleep schedule?
  - Prepare for any behavioral changes?
  - Avoid anything specific, such as any potential food-drug, alcohol-drug, drug-drug, or supplement-drug interactions?
    - **SIDENOTE:** be sure your healthcare team knows about every supplement that you take, no matter how safe or "over the counter" they are, they could impact certain the way drugs are absorbed.

# Master Appointment Schedule

Year: 2022



| Appointments           | JAN        | FEB        | MAR        | APR | MAY       | JUN        | JUL | AUG | SEP | OCT | NOV | DEC |
|------------------------|------------|------------|------------|-----|-----------|------------|-----|-----|-----|-----|-----|-----|
| Primary Care Physician |            | 2/15 @ 9am |            |     |           |            |     |     |     |     |     |     |
| Pediatrician           |            |            |            |     |           |            |     |     |     |     |     |     |
| Neurologist            |            |            | 3/3 @ 8am  |     |           |            |     |     |     |     |     |     |
| Neurosurgeon           |            |            |            |     |           |            |     |     |     |     |     |     |
| Cardiologist           |            |            |            |     |           |            |     |     |     |     |     |     |
| Nephrologist           |            |            |            |     |           |            |     |     |     |     |     |     |
| Dentist                |            |            |            |     |           | 6/1 @ 10am |     |     |     |     |     |     |
| MRI                    |            |            |            |     |           |            |     |     |     |     |     |     |
| EEG                    |            |            | 3/3 @ 9am  |     |           |            |     |     |     |     |     |     |
| EMU                    |            |            |            |     |           |            |     |     |     |     |     |     |
| EKG                    |            |            |            |     |           |            |     |     |     |     |     |     |
| Kidney Ultrasound/MRI  |            |            |            |     |           |            |     |     |     |     |     |     |
| Blood Draw             | 1/25 @ 8am |            | 3/15 @ 8am |     | 5/4 @ 8am |            |     |     |     |     |     |     |

# Doctor Visit Log



Date: 5/21/20 Doctor: Dr. Kojic Phone: 555-123-4567

Summary: Recent scans were unchanged from a tear ago. Will push MRI's to every 2 years. Follow-up?  Y  N

Date: \_\_\_\_\_

Date: \_\_\_\_\_ Doctor: \_\_\_\_\_ Phone: \_\_\_\_\_

Summary: \_\_\_\_\_ Follow-up?  Y  N

Date: \_\_\_\_\_

Date: \_\_\_\_\_ Doctor: \_\_\_\_\_ Phone: \_\_\_\_\_

Summary: \_\_\_\_\_ Follow-up?  Y  N

Date: \_\_\_\_\_

Date: \_\_\_\_\_ Doctor: \_\_\_\_\_ Phone: \_\_\_\_\_

Summary: \_\_\_\_\_ Follow-up?  Y  N

Date: \_\_\_\_\_



## Communication Log

**Date:** 10/21/21      **Name:** Mrs. Fracassa      **Type (phone, email):** phone

**Notes:** Called to update me on Logan reaching his IEP math and reading goals early. Wants to increase goals  
and will send over an amendment to his current IEP.

**Date:** \_\_\_\_\_      **Name:** \_\_\_\_\_      **Type (phone, email):** \_\_\_\_\_

**Notes:** \_\_\_\_\_

**Date:** \_\_\_\_\_      **Name:** \_\_\_\_\_      **Type (phone, email):** \_\_\_\_\_

**Notes:** \_\_\_\_\_

**Date:** \_\_\_\_\_      **Name:** \_\_\_\_\_      **Type (phone, email):** \_\_\_\_\_

**Notes:** \_\_\_\_\_









**NORD**<sup>®</sup>  
National Organization  
for Rare Disorders

## TUBEROUS SCLEROSIS COMPLEX TRAVEL & LODGING ASSISTANCE PROGRAMS

TRAVEL &  
LODGING  
ASSISTANCE

### What is the purpose of these programs?

These Programs provide patients diagnosed with tuberous sclerosis complex 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.



NORD provides assistance through the NORD TSC Clinical Trial Travel & Lodging Assistance Program and the NORD TSC Centers of Excellence Travel & Lodging Assistance Program. Both provide financial support and concierge travel and lodging arrangements for patients diagnosed with tuberous sclerosis complex.



### About the Programs:

The **NORD TSC Clinical Trial Travel & Lodging Assistance Program** provides financial assistance for travel and lodging expenses related to participation in a tuberous sclerosis complex trial within the United States.

The **NORD TSC Centers of Excellence Travel & Lodging Assistance Program** provides financial assistance for travel and lodging in furtherance of treatment for tuberous sclerosis complex at a TSC Alliance Center of Excellence or recognized TSC clinic.

Alone we are rare. Together we are strong.<sup>®</sup>

## What kinds of assistance are available?

- Airfare is available for patient and 1 caregiver with a 3-hour or greater driving distance from study or treatment site. Travel arrangements are scheduled and prepaid by NORD.
- Hotel is available for participants with a 3-hour or greater driving distance from study or treatment site. Hotel stay will be booked and prepaid by NORD (not to exceed \$200 plus tax/night).
- Ground transportation between the airport and hotel and/or to the study site will also be a covered expense up to \$50/trip.
- Hospital/clinic parking up to \$50/trip will be reimbursed with the submission of receipt.
- Mileage reimbursement at the current IRS rate will be reimbursed for those participants who are traveling by car and not utilizing airfare assistance.

## Is there a limit to the financial awards available in these program?

- Assistance to participant cannot exceed \$2,500 in a calendar year.

## How do I get more information and apply?

Contact NORD T&L Assistance Program

Monday-Friday 8:30 am – 6:00 pm ET

☎ 203.616.4320

📠 203.349.3199

✉ TSCTravel@rarediseases.org

✉ US MAIL to: NORD  
Attention: TSC Travel Program  
55 Kenosia Avenue  
Danbury, CT 06810

## NORD is Here for You

*NORD, a 501(c)(3) organization, is a patient advocacy organization dedicated to individuals with rare diseases and the organizations that serve them. NORD, along with its more than 300 patient organization members, is committed to the identification, treatment, and cure of rare disorders through programs of education, advocacy, research, and patient services.*

*NORD was founded by families struggling to obtain access to treatments and whose advocacy for change led to the passage of the Orphan Drug Act in 1983. NORD assists eligible patients (those with medical and financial needs) in affording the treatments and medical services their healthcare professionals have prescribed.*

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[rarediseases.org](http://rarediseases.org)

## Section Two: Navigating the medical journey

### Understanding the diagnosis

TSC is a genetic disorder that causes tumors to form in many different organs, primarily in the brain, eyes, heart, kidney, skin and lungs. Most people with TSC will live a normal life span. Although there is no cure, there is hope. Research has proven that early diagnosis and intervention(s) are key for optimizing long-term outcomes. Advancements in research continue to deliver new and improved therapeutic options. The TSC journey is unique to each individual and family – even siblings and identical twins can have different experiences. Understanding the disease will give you hope, strength, and empowerment through all life stages.

After allowing time to process the initial diagnosis of TSC, you can start navigating options with your healthcare team to achieve your treatment goals. Understanding all your treatment options at every stage of the journey is essential. Our TSC Support Navigators can help you identify treatment options and clinical trials that may be right for you.

As previously mentioned, another way to get informed is with TSC Academy, an online educational platform created for the TSC community. This resource features lessons about the basic, preclinical, and clinical science of TSC, how research helps us understand and treat the disease, how the TSC Alliance; federal, state, and local governments; and other stakeholders work together to find a cure and how you play a role.

### Clinical manifestations

Those with TSC can experience a variety of organ-specific manifestations. Some of these manifestations are present at birth (or during fetal development) whereas others manifest over the lifespan. Because genetic testing only captures 85-90% of those with an identified mutation, clinical diagnostic criteria help clinicians identify major and minor features that are considered “hallmark” findings of TSC. In 2021, the clinical diagnostic criteria re updated and are reflected below. Not all features of TSC are listed, but not all ailments are associated with TSC; therefore, it's important to have ongoing discussions with your TSC expert provider so that proper treatment recommendations can be initiated or referral to other specialists is not delayed.

### 2021 TSC diagnostic criteria updates

The International TSC Consensus Group was established in 2012 to update recommendations from the 1998 International Consensus Conference. Due to the advancements of research and understanding of the disease, leaders of the working group collaborated in 2018 at the World TSC Conference in Dallas, Texas over two days to confirm or amend prior recommendations or provide new recommendations by each of the organ-specific working groups. These updated recommendations were published in 2021. These recommendations provide clinicians with peer-reviewed and expert-driven recommendations so any clinician worldwide can assist in providing quality surveillance and management to those affected. The full text recommendations can be found on the TSC website under Healthcare Professionals/Key Medical Publications, but for those of you who would like a general overview please reference the Surveillance and Management Recommendations for TSC in the appendix of this section.

### TSC genetics

TSC affects multiple organs throughout the lifetime. Both the TSC1 and TSC2 genes hold the instructions for creating proteins called hamartin and tuberin, respectively. These proteins form a complex (essentially a protein sandwich) that works in a delicate biochemical pathway. This pathway is called the mTOR pathway, where mTOR stands for “mechanistic target of rapamycin.” The pathway carefully regulates cell growth in almost every cell type in the body. The TSC1 and TSC2 proteins, when functioning together properly, regulate a key step in this pathway and suppress tumor growth.

When either the TSC1 or TSC2 gene mutates, cell growth cannot be adequately controlled, which leads to TSC. Hamartin, tuberin, and mTOR are expressed in many different cells throughout the body, which explains why so many organs can be affected by TSC. However, researchers are still working diligently to determine why TSC manifests so differently between different people.

Tuberous sclerosis complex is a genetic disease that can be inherited from one parent with TSC or can result from a spontaneous genetic mutation. Children have a 50 percent chance of inheriting TSC if one of their parents has this condition. Researchers estimate that only one-third of TSC cases are known to be inherited. The other two-thirds result from a spontaneous and unpredictable mutation occurring during conception or very early development of the human embryo. To learn more about the mTOR pathway and genetics or for the Primer course, please go to the TSC Academy.

## TSC over time

For those who have been on the TSC journey since childhood and have successfully transitioned from pediatrics to adulthood, the following information is dedicated to discussing the manifestations of TSC that may or may not have been problematic during childhood or adolescence, but now take more center-stage to your medical journey. Building on the previous navigation guide and a basic understanding of clinical manifestations, this information is specific to the most common or problematic issues associated with adult years. If you are not familiar with the TSC Clinical Manifestations, you can preview them at the beginning of this section.

## Neurological manifestations

During infancy through young adulthood seizures and subependymal giant cell astrocytoma (SEGA) dominate much of your medical journey. At this point (>27 years of age), it is not as common to develop epilepsy if you have not already done so. SEGAs at this point typically do not grow or become problematic. However, if you or your adult child had ventricular enlargement due to a SEGA and have remained asymptomatic, you should continue with MRI imaging of the brain periodically, but not as frequent as it was during earlier in life where rapid growth and development were still occurring.

Seizures are still problematic for many and unfortunately several have developed refractory epilepsy that will remain challenging to manage indefinitely until we have better knowledge and resources to assist with this manifestation. EEGs will still be a common diagnostic approach routinely and for atypical changes such as changes in seizure type or frequency, change in behavior, unexplained sleepiness or vomiting. For those who were never able to achieve seizure control during their younger years, this is also a time where notable neurocognitive damage will be permanent if all interventions have failed. We will focus more on TSC-associated neuropsychiatric disorders (TAND) in Section Five. Interventions for neurocognitive deficits are specific to the need and navigating those barriers will be discussed throughout the remainder of the guide. For the most up-to-date information, you can go to the TSC Navigator.

## Seizure clusters and status epilepticus in TSC

Epilepsy is a very common manifestation of TSC, occurring in 84% of people registered in the TSC Alliance Natural History Database. Potential complications of epilepsy include seizure clusters and status epilepticus.

### What is a seizure cluster?

The definition of a seizure cluster varies from one individual to another, but in general the term refers to a group of seizures occurring over several hours or days in a way that differs from an individual's usual seizure pattern. For one person, having three seizures over the course of two hours may represent a seizure cluster, while for another a seizure cluster may be represented by the occurrence of three seizures over the course of two days.

Adverse effects of seizure clusters include risk of injury from the seizures themselves, prolonged recovery time after seizures and missed work, school or social activities. Seizure clusters may progress into status epilepticus.

### What is status epilepticus?

The official definition of status epilepticus has evolved over time, but the term generally refers to a prolonged seizure that may not stop on its own without intervention. The most recent definition presented by the International League Against Epilepsy identifies generalized convulsive (tonic-clonic) status epilepticus as a seizure lasting longer than five minutes, with an increased risk of brain injury if the seizure continues for 30 minutes or longer.

For other seizure types, such as focal or absence seizures, these time points may be more prolonged. The majority of seizures in individuals with TSC are short and terminate on their own in less than two to three minutes. However, if status epilepticus occurs, it is a medical emergency that may result in severe consequences if not treated rapidly.

### **What causes seizure clusters and status epilepticus?**

People with intractable epilepsy (seizures that are not controlled by medication) are at higher risk for having seizure clusters or status epilepticus, but these episodes may also occur in people whose seizures are usually well controlled.

Some common triggers for seizure clusters or status epilepticus include:

- illness,
- missed antiseizure medication doses,
- sleep deprivation, and
- stress.

For some women, seizure clusters may occur in relation to their menstrual cycle. However, seizure clusters or status epilepticus may also occur without an apparent trigger. Keeping a seizure diary may be helpful in identifying triggers.

### **How can seizure clusters and status epilepticus be treated?**

Prompt treatment of seizure clusters and status epilepticus is important in limiting the duration of seizures and may prevent the need for more intensive hospital-based treatment. Medications to treat seizure clusters or status epilepticus are often called rescue medications. Several rescue medications are now available for out-of-hospital treatment. All these medications are part of the benzodiazepine group of drugs, which are the first choice for treatment of prolonged seizures in the hospital. While benzodiazepines used in the hospital are usually given intravenously (IV), benzodiazepine medications available for out-of-hospital treatment may be given orally, rectally or nasally.

All benzodiazepines have the same potential side effects, which include drowsiness, dizziness, and impaired coordination. At high doses they may cause respiratory depression. In general, emergency services should be contacted after administration of any of these medications to provide assistance if the seizure continues or if serious adverse effects occur.

1. **Oral medications** used for seizure clusters include clonazepam (Klonopin®), lorazepam (Ativan®), and diazepam (Valium®). These medications may be swallowed or given by gastrostomy tube, and some come in forms that can dissolve on the tongue. Absorption of medications given by mouth is slower than the other treatment options, and these are therefore not a good option for treating status epilepticus. It is important that a person be alert and responsive before administering an oral medication to reduce the risk of aspiration. A person experiencing a seizure may also inadvertently bite down and injure someone trying to put something in his or her mouth.
2. A **rectal medication** used for seizure clusters or status epilepticus is diazepam gel (Diastat®). Diastat is provided as a needleless syringe containing a weight-based dose of diazepam gel that is administered into the rectum, where it is quickly absorbed into the bloodstream. Diastat is often preferred for use in young children, as the method of administration is undesirable for many adolescents and adults. The medication does not require refrigeration and has a long shelf life. Diastat is FDA-approved for the treatment of children over two years of age and adults, but many pediatric neurologists also recommend the use of this medication in children under two years of age.
3. **Nasal medications** may be used to treat seizure clusters but are not FDA-approved for the treatment of status epilepticus. These medications are sprays that are administered into the nose. There are currently two FDA-approved nasal medications: intranasal midazolam (Nayzilam®) for ages 12-plus and intranasal diazepam (Valtoco®) for ages 6-plus. Both these options are safe to store at room temperature and have a long shelf life. There are currently no FDA-approved intranasal rescue medications for children younger than 6 years of age. There are several potential benefits to nasal medications, including ease of usage, safety for the person administering the medication, and avoidance of potential embarrassment for adolescents and adults that may otherwise require a rectal medication. However, nasal medications may not be a good option for everyone. There are limits to how frequently these medications should be given, so they may not be a good option for those with frequent seizure clusters.

### **When should rescue medications be used for seizure clusters or status epilepticus?**

Every person with TSC and epilepsy should develop a plan with his or her medical provider that details the indications for use of a seizure rescue medication and which medication should be used. This plan should be implemented as soon as possible once a seizure cluster or status epilepticus is identified. The plan should be written down and be available for anyone who might be with the individual when he or she experiences a seizure. For those known to have frequent seizure clusters or status epilepticus, a plan should also be available for emergency medical personnel. Most schools and daycare providers require a written Seizure Action Plan for all children with epilepsy that details the types of seizures they have, what should be done if they have a seizure, and when rescue medications should be given.

### **Kidney manifestations**

Kidney (renal) manifestations such as cystic disease and renal angiomyolipomas continue to grow throughout this part of the lifespan. Not only are you/your loved one aging, in which chronic kidney disease tends to manifest in general adulthood, but the progression of TSC-specific manifestations and long-term medication use all contribute to factors that place you or your loved one at risk for chronic kidney disease and/or hypertension. Surveillance with MRI of the abdomen is still recommended every 1 to 3 years during this timeframe. The goal is to be preventative and proactive to decrease progressing to end-stage renal disease, which will be covered later in this section. Ideal diet, daily exercise and maintaining an optimal blood pressure are essential for not only general and cardiovascular health purposes but also to minimize declining renal function.

### **End-stage renal disease**

End-stage renal disease (ESRD) is when the kidneys permanently stop working and require long-term dialysis or a kidney transplant to maintain life. While ESRD is rare in TSC, chronic kidney disease is not. Chronic kidney disease takes several decades to decline to the point of ESRD. Those with TSC and polycystic kidney disease (PKD) are at a much higher risk of kidney failure over their lifespan. Proper kidney care is essential to preserve renal function. Great ways to proactively protect your kidneys include careful blood pressure management, prevention of protein in the urine (proteinuria) and a healthy diet. Over time, interventions like reoccurring embolization, nephrectomies, renal hemorrhages and acute illnesses can take a toll on kidney function. If you find yourself in a situation and your healthcare provider starts to discuss dialysis or renal transplant in your future, the National Kidney Foundation and PKD Foundation have excellent resources to help you make more informed decisions.

### **Lung manifestations**

Individuals with TSC are at a higher risk of developing lymphangiomyomatosis (LAM). LAM is a rare lung disease that affects women more often than men, usually between the onset of puberty and menopause. It can cause shortness of breath or other complications. Treatments for LAM can include medication (Rapamune®, sirolimus), oxygen therapy or lung transplantation in very advanced diseases. Ongoing screening and management are recommended throughout the lifespan. You or your loved one can continue to make conscious choices such as smoke cessation, minimize occupational exposures and avoid routine use of exogenous estrogen (birth control) or doxycycline for the treatment of LAM. Diagnostics testing and imaging should be utilized every 1 to 3 years or more frequent pending your unique situation. Proactively understanding symptoms that you should report to your clinical team would include connective tissues disease symptoms, chyle leak, dyspnea, cough or experiencing spontaneous pneumothorax. Should you or your loved one have these symptoms and are not explained by other factors, you should notify your TSC specialist for further recommendations. The LAM Foundation offers excellent resources and support for those looking for additional information on LAM.

### **Heart manifestations**

Typically during adulthood, the common pediatric manifestation of cardiac rhabdomyomas has completely regressed. However, some individuals with TSC will continue to have small evidence of these lesions throughout adulthood. There is not sufficient evidence to know if these lesions cause issues during adulthood; however, one problematic issue that can occur is irregular heartbeats known as cardiac arrhythmias or conduction defects. One arrhythmia that has been well documented within the literature is Wolfe-Parkinson-White Syndrome (WPW Syndrome). Because of these potential possibilities diagnostic testing such as echocardiogram (ECHO) and electrocardiogram (EKG/ECG) should be completed every 1 – 3 years for those who do have symptoms. Those who do will most likely have more frequent follow-up and diagnostic surveillance that your cardiologist feels necessary. There are no specific TSC guidelines for these manifestations, which are managed the same as the general public experiencing these same symptoms.

## **Skin, eyes and teeth manifestations**

Most of these manifestations do not cause life-altering problems. However, facial angiofibromas do continue to grow throughout this part of the lifespan and can become painful and cause recurrent bleeding concerns and rarely, nasal airway obstruction. First-line treatment recommendation focuses on mTOR inhibitor treatment. In 2022, the FDA approved the first topical rapamycin gel for those diagnosed with facial angiofibromas. Ungual fibromas (lesions that grow in fingernails or toenails) can also increase during adulthood. If those become problematic or bothersome, surgical removal can be completed by a dermatologist. Currently, there is not sufficient research or literature to support retinal changes that occur in the aging adult with TSC. Therefore, it is recommended to continue to have annual ophthalmic evaluations to assess for visual impairments. If you or your loved one experience a sudden loss of vision of any sort, double vision, blurry vision or a sudden onset of significantly elevated blood pressure, you should notify your local medical team immediately. As with the eyes, teeth manifestations are another area that has not received much longitudinal surveillance from a research standpoint. We do know enamel pits remain an issue throughout the lifespan, so having your dentist preventively utilize sealants to minimize tooth decay is appropriate every 6 months. It is important to know oral fibromas can occur near the teeth or within the bony jaw, and these should be treated with surgical excision or curettage when present.

## **Other manifestations**

Many individuals with TSC have been identified with other organ specific lesions such as functional and nonfunctional pancreatic neuroendocrine tumors (PNETS). Other locations include the GI track (bowel) and liver (hepatic lesions). Typically, these are benign; however, depending on the size and location, your local provider might refer you to a gastroenterology specialist or oncologist for further recommendations. It is important that you articulate up front you or your loved one has TSC. If your TSC expert is not aware, you should notify him or her about the finding so he/she can collaborate with the other medical specialists in providing the most current and up-to-date information regarding these findings.

## **Treatment pathways (surgical evaluations, emerging medications and recent breakthroughs)**

Treatment options directly depend on the manifestation, such as the type of seizure or the organ system that is causing an issue. If your child presented with seizures, the most critical priority is gaining seizure control. All efforts should be made to reduce the total number of daily seizures. Not all individuals with TSC have the same form of seizures, so one approach does not work for everyone. It is helpful to remember when making shared decisions with your medical team to discuss the best approach for your loved one.

Other treatment options to consider are to address high blood pressure (hypertension) or proteinuria (protein in urine), which can be common to individuals who experience renal (kidney) involvement. If your child has been diagnosed with continuous gene syndrome, also known as TSC-Polycystic Kidney Disease (PKD), vigilant blood pressure control and urinalysis are important to observe and maintain kidney function over time. There are many options for blood pressure and proteinuria management; therefore, if your child has cystic involvement, it is essential to have close followup with a nephrologist who can best proactively manage these manifestations.

## **FDA-approved treatments/medications for TSC**

Advances in research continue to deliver new and improved therapeutic options. Five FDA-approved drugs with a TSC-specific indication are available, but there are also approved treatments for manifestations of TSC, like seizures, that many TSC individuals rely on. These include Afinitor® (everolimus), Rapamune® (sirolimus), Sabril® and Vigadrone® (vigabatrin), Epidiolex® (cannabidiol) and Hyftor® (topical sirolimus).



# 2021 TUBEROUS SCLEROSIS COMPLEX

## DIAGNOSTIC CRITERIA, SURVEILLANCE AND MANAGEMENT RECOMMENDATIONS

### Diagnostic criteria

| Major Criteria  | Minor Criteria                  |
|---|---------------------------------|
| Hypomelanotic macules ( $\geq 3$ ; at least 5mm diameter) | "Confetti" skin lesions         |
| Angiofibroma ( $\geq 3$ ) or fibrous cephalic plaque      | Dental enamel pits ( $\geq 3$ ) |
| Ungual fibromas ( $\geq 2$ )                              | Intraoral fibromas ( $\geq 2$ ) |
| Shagreen patch  | Retinal achromic patch          |
| Multiple retinal hamartomas                               | Multiple renal cysts            |
| Multiple cortical tubers and/or radial migration lines*   | Nonrenal hamartomas             |
| Subependymal nodule ( $\geq 2$ )                          | Sclerotic bone lesions          |
| Subependymal giant cell astrocytoma                       |                                 |
| Cardiac rhabdomyoma                                       |                                 |
| Lymphangiomyomatosis (LAM)**                              |                                 |
| Angiomyolipomas ( $\geq 2$ )**                            |                                 |

**Definite TSC:** 2 major features or 1 major feature with 2 minor features.

**Possible TSC:** Either 1 major feature or  $\geq 2$  minor features.

\*Includes tubers and cerebral white matter radial migration lines.

\*\*A combination of the 2 Major clinical features LAM and angiomyolipomas without other features does not meet criteria for a definite diagnosis.

**Genetic diagnosis:** A pathogenic variant in TSC1 or TSC2 is diagnostic for TSC. Most TSC-causing variants are sequence variants that clearly prevent TSC1 or TSC2 protein production. Some variants compatible with protein production (e.g., some missense changes) are well established as disease-causing. Other variant types should be considered with caution.

## Surveillance and management recommendations for newly diagnosed or suspected tuberous sclerosis complex (TSC)

| Organ System or Specialty Area | Recommendations  |   |
|--------------------------------|--|---|
| <b>GENETICS</b>                | <p>Obtain three-generation family history to assess for additional family members at risk of TSC.</p> <p>Offer genetic testing for family counseling or when TSC diagnosis is in question but cannot be clinically confirmed.</p>  |    |
| <b>BRAIN</b>                   | <p>Obtain magnetic resonance imaging (MRI) of the brain to assess for the presence of tubers, subependymal nodules (SEN), migrational defects, and subependymal giant cell astrocytoma (SEGA).</p> <p>During infancy, educate parents to recognize infantile spasms and focal seizures, even if none have occurred at the time of first diagnosis.</p> <p>Obtain baseline routine electroencephalogram (EEG) while awake and asleep. If abnormal, especially if features of TSC-associated neuropsychiatric disorders (TAND) are also present, follow up with 8- to 24-hour video EEG to assess for seizure activity.</p>  |    |
| <b>TAND</b>                    | <p>Perform comprehensive assessment for TSC-associated neuropsychiatric disorders (TAND) across all levels of potential TAND manifestations.</p> <p>Refer as appropriate to suitable professionals to initiate evidence-based interventions based on the TAND profile of needs identified above.</p> <p>Provide parent/caregiver education and training about TAND to ensure families know what to look out for in emerging TAND manifestations (e.g. autism spectrum disorder, language disorders, attention deficit hyperactivity disorder, anxiety disorders).</p> <p>Provide psychological and social support to families around diagnosis, coming to terms with the diagnosis of TSC and TAND, and ensure strategies are in place to support caregiver wellbeing.</p> |    |
| <b>KIDNEY</b>                  | <p>Obtain MRI of the abdomen to assess for the presence of angiomyolipomas and renal cysts.</p> <p>Screen for hypertension by obtaining an accurate blood pressure.</p> <p>Evaluate renal function by determination of glomerular filtration rate (GFR).</p>   |  |
| <b>LUNG</b>                    | <p>Inquire about tobacco exposure, connective tissue disease manifestations, signs of chyle leak, and pulmonary manifestations of dyspnea, cough, and spontaneous pneumothorax in all adult patients with TSC.</p> <p>Perform baseline chest CT in all females, and symptomatic males, starting at the age of 18 years or older.</p> <p>Perform baseline PFTs and 6MWT in patients with evidence of cystic lung disease consistent with LAM on the screening chest CT.</p>   |  |
| <b>SKIN</b>                    | <p>Perform a detailed clinical dermatologic inspection/exam.</p>   |  |
| <b>TEETH</b>                   | <p>Perform a detailed clinical dental inspection/exam.</p>   |  |
| <b>HEART</b>                   | <p>Consider fetal echocardiography to detect individuals with high risk of heart failure after delivery when rhabdomyomas are identified via prenatal ultrasound.</p> <p>Obtain an echocardiogram in pediatric patients, especially if younger than three years of age.</p> <p>Obtain an electrocardiogram in all ages to assess for underlying conduction defects.</p>  |  |
| <b>EYE</b>                     | <p>Perform a complete ophthalmologic evaluation, including dilated funduscopy, to assess for retinal findings (astrocytic hamartoma and achromic patch) and visual field deficits.</p>   |  |

## Surveillance and management recommendations for patients already diagnosed with definite or possible TSC

| Organ System or Specialty Area | Recommendations   |
|--------------------------------|---|
| GENETICS                       | <p>Offer genetic testing and family counseling if not performed previously.</p>    |
| BRAIN                          | <p>Obtain magnetic resonance imaging (MRI) of the brain every 1 to 3 years in asymptomatic TSC patients younger than age 25 years to monitor for new occurrence of subependymal giant cell astrocytoma (SEGA). Patients with large or growing SEGA, or with SEGA causing ventricular enlargement but yet are still asymptomatic, should undergo MRI scans more frequently, and the patients and their families should be educated regarding the potential of new symptoms. Patients with asymptomatic SEGA in childhood should continue to be imaged periodically as adults to ensure there is no growth.</p> <p>Surgical resection should be performed for acutely symptomatic SEGA. Cerebral spinal fluid diversion (shunt) may also be necessary. Either surgical resection or medical treatment with mechanistic target of rapamycin inhibitors (mTORi) may be used for growing but otherwise asymptomatic SEGA. For large tumors, if clinical condition enables, neoadjuvant treatment with mTORi may facilitate surgery. Minimally invasive surgical techniques may increase surgical safety in selected patients. In determining the best treatment option, discussion of the complication risks, adverse effects, cost, length of treatment, and potential impact on TSC-associated comorbidities should be included in the decision-making process.</p> <p>Obtain routine electroencephalogram (EEG) in asymptomatic infants with TSC every 6 weeks up to age 12 months and every 3 months up to age 24 months, as abnormal EEG frequently precedes onset of clinical seizures.</p> <p>Obtain routine EEG in individuals with known or suspected seizure activity. The frequency of routine EEG should be determined by clinical need rather than a specific defined interval. Prolonged video EEG, 24 hours or longer, is appropriate when seizure occurrence is unclear or when unexplained sleep, behavioral changes, or other alteration in cognitive or neurological function is present.</p> <p>Vigabatrin is the recommended first-line therapy for infantile spasms. Adrenocorticotropic hormone (ACTH), synthetic ACTH or prednisolone can be used if treatment with full-dose vigabatrin for 2 weeks has not correlated with clinical and EEG improvement.</p> <p>Antiseizure medications (ASM) for other seizure types in TSC should generally follow that of other epilepsies. Everolimus and a specific cannabidiol formulation are approved by regulatory authorities for treatment of seizures associated with TSC. No comparative effectiveness data exist to recommend ASM, everolimus, cannabidiol, or dietary therapies over one another in specific subsets of patients.</p> <p>Epilepsy surgery should be considered for medically refractory TSC patients at epilepsy surgery centers with expertise in TSC. Special consideration should be given to children at younger ages experiencing neurological regression and evaluation for surgery should be performed at epilepsy surgery centers with experience and expertise in TSC.</p>  |
| TAND                           | <p>Perform annual screening for TAND, using validated screening tools such as the TAND Checklist (<a href="http://tandconsortium.org/checklists/">tandconsortium.org/checklists/</a>). Screening may be done more frequently depending on clinical needs. When any concerns are identified on screening, proceed to further evaluations by appropriate professionals to diagnose and treat the relevant TAND manifestation(s).</p> <p>Perform comprehensive formal evaluation for TAND across all levels of TAND at key developmental time points: infancy (0-3 years), preschool (3-6 years), pre-middle school (6-9 years), adolescence (12-16 years), early adulthood (18-25 years), and as needed thereafter.</p> <p>Refer to appropriate professionals for the management/intervention of relevant TAND manifestations. Interventions should be personalized to the TAND profile of each individual and be based on evidence-based practice guidelines/practice parameters for individual manifestations (e.g. autism spectrum disorder, attention deficit hyperactivity disorder, anxiety disorder).</p> <p>Aim for early identification of TAND manifestations and early intervention.</p> <p>Many people with TSC have academic/scholastic difficulties. Therefore, always consider the need for an individual educational program (IEP/IEDP).</p> <p>Sudden and unexpected change in behavior should prompt physical evaluation to look at potential medical causes (e.g., SEGA, seizures, renal disease, medications).</p> <p>Provide psychological and social support to families and caregivers and ensure strategies are in place to support caregiver wellbeing. Continue to provide parent/caregiver education and training about TAND to ensure families know what to look out for in emerging TAND manifestations across the lifespan.</p>    |

|                         |   |   |
|-------------------------|---|---|
| <p><b>RENAL</b></p>     | <p>Obtain MRI of the abdomen to assess for the progression of angiomyolipoma and renal cystic disease every 1 to 3 years throughout the patient's lifetime.</p> <p>Assess renal function including determination of glomerular filtration rate and blood pressure at least annually.</p> <p>Embolization followed by corticosteroids is first-line therapy for angiomyolipoma presenting with acute hemorrhage. Nephrectomy is to be avoided. For asymptomatic, growing angiomyolipoma measuring larger than 3 cm in diameter, treatment with an mTOR inhibitor is the recommended first-line therapy. Selective embolization or kidney-sparing resection are acceptable second-line therapy for asymptomatic angiomyolipoma.</p>   |    |
| <p><b>PULMONARY</b></p> | <p>Inquire about smoking, occupational exposures, connective tissue disease (CTD) symptoms, chyle leak, and pulmonary manifestations such as dyspnea, cough, and spontaneous pneumothorax in all adult patients at each clinic visit.</p> <p>For adult females with a negative screening CT who remain asymptomatic, obtain high resolution CT (HRCT) to screen for the presence of LAM every 5 years through menopause. Low-dose CT protocols preferred.</p> <p>For patients with evidence of cystic lung disease consistent with LAM on screening CT, obtain follow-up HRCT after 1 to 3 years, and on a case-by-case basis thereafter at least every 5 years depending upon the individual circumstances. Low-dose CT protocols preferred.</p> <p>Perform routine serial PFT monitoring at least annually in patients with evidence of LAM on HRCT and more frequently in patients who are progressing rapidly or who are being monitored for response to therapy.</p> <p>Use mTOR inhibitors for treatment of LAM in patients with abnormal lung function (FEV1 &lt; 70% predicted), physiological evidence of substantial disease burden (abnormal DLCO (&lt;80% or less than lower limit of normal [when available]), air trapping (RV &gt; 120%), resting or exercise-induced oxygen desaturation, rapid decline (rate of decline in FEV1 &gt; 90ml/year), and problematic chylous effusions.</p> <p>Counsel patients regarding the risk of pregnancy and exogenous estrogen use. Avoid routine use of hormonal therapy or doxycycline for the treatment of LAM. Advise patients against tobacco smoke exposure.</p> <p>Trial inhaled bronchodilators in patients with symptoms of wheezing, dyspnea, chest tightness, or obstructive defect on spirometry, with continued use in patients who derive symptomatic benefit.</p> <p>Consider measurement of annual VEGF-D levels in patients who are unable to perform reliable PFTs to monitor adequacy of pharmacodynamic suppression of the mTOR pathway.</p> |    |
| <p><b>SKIN</b></p>      | <p>Perform annual skin examinations for children with TSC. Adult dermatologic evaluation frequency depends on the cutaneous manifestation. Close surveillance and intervention are generally recommended for TSC-related skin lesions that rapidly change in size and/or number, cause functional interference, pain, or bleeding, or inhibit social interactions.</p> <p>Provide ongoing education on sun protection.</p> <p>For flat or minimally elevated lesions, topical mTOR inhibitor treatment is recommended. Watch for improvement in skin lesions over several months; if lesions do not improve, or if earlier intervention is indicated, then consider use of surgical approaches. For protuberant lesions, consider surgical approaches (e.g. excision, lasers).</p>  |   |
| <p><b>TEETH</b></p>     | <p>Perform a detailed clinical dental inspection/exam at minimum every 6 months. Take a panoramic radiograph to evaluate dental development or if asymmetry, asymptomatic swelling, or delayed/abnormal tooth eruption occurs. Enamel pits may be managed by preventive measures as first-line treatment (sealants, fluoride). They may be managed by restorations if preventive measures fail, or if symptomatic, carious, or there is an aesthetic concern. Symptomatic or deforming oral fibromas and bony jaw lesions should be treated with surgical excision or curettage when present.</p>   |  |
| <p><b>HEART</b></p>     | <p>Obtain an echocardiogram every 1 to 3 years in asymptomatic pediatric patients until regression of cardiac rhabdomyomas is documented. More frequent or advanced diagnostic assessment may be required for symptomatic patients.</p> <p>Obtain electrocardiogram every 3 to 5 years in asymptomatic patients of all ages to monitor for conduction defects. More frequent or advanced diagnostic assessment such as ambulatory and event monitoring may be required for symptomatic patients.</p>  |  |
| <p><b>EYE</b></p>       | <p>Perform annual ophthalmic evaluation for those with or without visual symptoms at baseline. Rare cases of aggressive lesions or those causing vision loss due to their location affecting the fovea or optic nerve may require intervention. mTOR inhibitors have been used with some success to treat problematic retinal astrocytic hamartomas.</p> <p>For patients receiving vigabatrin, there are specific concerns related to visual field loss which appears to correlate with total cumulative dose. Physicians responsible for monitoring children on vigabatrin can offer serial fundus examinations to detect retinal changes.</p>   |  |
| <p><b>OTHER:</b></p>    | <p>Identification of unexpected functional and nonfunctional pancreatic neuroendocrine tumors (PNETS) have been found during abdominal MRI surveillance in individuals with TSC. Further monitoring and evaluation should be referred to endocrinology.</p>   |   |

## Section Three: Medical, prescription and and insurance challenges

### Medical challenges

Medical challenges do not just apply to the medical condition. A complex medical condition like TSC, which often requires multiple medications and/or treatments, also has additional challenges and barriers, such as primary insurance, secondary insurance, Medicaid, waiver, and access to medication.

### Pharmacy coverage

Prescription drug plans are part of all commercial and government insurance plans in the United States; however, each plan is different per corporation or organization and state. When starting to understand your coverage you should immediately get familiar with your plan's prescription drug list known as a PDL. This list will not only show which medications will be covered under your plan (called a formulary), but what category or "tier" each of these medications are listed under. Tiers represent a cost level. There are four tiers to a formulary list, tier 1 being the lowest cost and tier 4 being the highest cost.

Tiers 1 to 3 usually require some co-pay, and it is not uncommon to have your co-pay cost listed on your pharmacy drug card. Most of these medications are generic, and brand name medications are often higher up on the tier list. If you have Medicaid or Medicare, it is not uncommon for your insurance to cover brand names. Some states have restrictions on how many brand-named drugs you can use per month (including antibiotics). Tier 4, also known as the "specialty tier," is where most specialty medications for rare diseases and new epilepsy medications are found.

Cost-sharing is the major difference between the specialty tier and those preceding it. While for lower tier medications you'd typically be charged a flat co-payment for your prescription, specialty tier medicines usually come with a coinsurance payment. Coinsurance is a percentage-based rate – so if your coinsurance payment rate is 20%, you'll find yourself paying for 20% of the medication's overall retail cost. Because some medications can cost several thousands of dollars per month, you may end up meeting your insurance deductible very quickly just on coinsurance payments alone. Once you've met your annual deductible, insurers usually begin covering more (up to 80% to 100%) of medication costs. This is why it is very important to understand your deductible plan, because each year in January, your progress toward "meeting your deductible" (i.e., the amount of money you spend each year) resets.

Many people utilize co-pay assistance cards so medications can be affordable; however, under the current co-pay accumulator programs, this approach can end up costing you more money in the long run. The TSC Alliance is advocating for co-pay accumulator programs to be removed. For more information on prescription process, including quick videos on these important topics check out the Alliance for Patient Access (AfPA), listed in the appendix section, which advocates for patient access to approved therapies and appropriate clinical care.

Another important aspect of pharmacy coverage is step therapy. This is also called "fail first" policy that was developed by insurers to help control cost. This means some plans require you to try cheaper and outdated drugs FIRST before they will cover a specific drug prescribed by your healthcare provider. This is a very frustrating process and can be risky for those with a rare disease, like TSC, and epilepsy. As you are understanding your pharmacy benefits, pay attention and ask which medications require step therapy. It is equally important to understand that many other advocacy groups have taken a proactive stance on getting step therapy legislation changed. For more insight, [Steptherapy.com](http://Steptherapy.com) is a great resource to find out if there is an override request available in your state and what to do if your override request was denied.

## Retail pharmacy vs. specialty pharmacy

There are two main types of pharmacies most individuals utilize, retail and specialty. The main differences between the two are the type of medication they can dispense and the services offered. Retail pharmacies include your local Walgreens, CVS, or Wal-Mart. Common prescriptions (such as antibiotics, blood pressure and allergy medications) are usually sent to a retail pharmacy and, if you're lucky, you can pick them up after you've done some grocery shopping.

Certain treatments for rare diseases require specialized pharmacists who are knowledgeable about medications for complex diseases. This includes capabilities for researching financial assistance options or side effects, and constant communication with the prescribing provider's office. A specialty pharmacy can either be a local specialty pharmacy or a mail order pharmacy selected by your insurance provider.

While there are many moving pieces, you can work proactively to make obtaining prescriptions as seamless as possible. Ensure all stakeholders have up-to-date, accurate information – your insurance card, phone number, and mailing address are critical to confirm at each encounter. At each visit with your healthcare team, be sure to confirm they have the correct pharmacy on file to ensure your prescriptions go to the right place. This is especially important if you have multiple prescriptions sent to different pharmacies (e.g., a retail pharmacy handling a standard prescription and a specialty pharmacy handling another).

## Connecting with your pharmacist

Your specialty pharmacist is an important member of your extended healthcare team. When you are first connected with a specialty pharmacy, ask for a direct number. Introduce yourself to the pharmacist and get his or her name since you may be able to be redirected to the same person in the future should you need to troubleshoot any problems.

Pharmacists can also help you understand important aspects of your prescription. For example, they can identify the tier for the medication and advise of any continual or renewing requirements (such as a prior authorization). Furthermore, if medications require bloodwork or additional paperwork, the pharmacist can guide you to what and how frequently it is needed per your plan's policy.

A specialty pharmacy will try to communicate with your prescribing provider's office via fax to obtain records such as bloodwork or prior authorizations. Unfortunately, often faxes from specialty pharmacies may be sent to a centralized fax hub within your healthcare institution and can be lost in the void of paperwork. Remember, you are an important conduit between the specialty pharmacy and your prescribing provider – connect them to each other by providing the specialty pharmacy your provider's office line or direct fax line, and be sure to request copies of important correspondence in case they get lost in the shuffle. You should proactively call the provider's office if the pharmacy needs anything from them to prevent delay in receiving the medication. The provider's office can also expedite the process from their end too.

## Prior authorization issues

Medications prescribed for TSC-related manifestations often require a prior authorization (PA). It is important to know which medications will require a PA, and how long the PA is approved for each medication. For medications that are controlled substances or non-preferred medications, it is common that a new PA cannot be initiated until four days from the last dose covered by the preceding PA. Therefore, it is essential to know exactly what your healthcare team will need to have to initiate the new PA before you run out of medication. Medications requiring a PA will also be listed on your pharmacy drug list (PDL). For those who need an example letter, please check out the Prior Authorization Issues in the TSC Navigator for the most current templates.

## Insurance barriers

One of the most common insurance barriers is getting out-of-state healthcare visits covered. Because of the challenges associated with out-of-state healthcare, it is important to have a local team (pediatrician or internist and, if needed, a neurologist) who can help provide stability in your care. Here are some helpful tips on this process:

- The individual being seen **MUST** have an established primary care provider (who is licensed in the state the individual lives in) who sees them routinely. This primary care provider needs to have seen the individual at least once within the current calendar year **BEFORE** an out-of-state referral is made. Medicaid coverage for out-of-state referrals is often delayed because:
  - o Your primary care office staff typically need 30 days to request records and be able to review.
  - o Your primary care office staff must justify to your home state's Medicaid why services need to be rendered **OUTSIDE** the state, and they cannot do so without making a complete evaluation of the patient's records.
- Ensure insurance information is up to date. This needs to be confirmed **YEARLY** in January by the primary caregiver/parent, especially if multiple insurances are involved. Most require annual submission of forms, so insurance does not lapse. Not knowing and having them "termed" or "inactive" will always result in **DELAYED** care.
- When referred to out of state care with Medicaid, ensure **ACCURATE** information is provided and the out-of-state provider is enrolled with that state's Medicaid program.
  - o Contact the out-of-state clinic prior to the referral being placed to ensure the provider(s) are enrolled.
    - **IF NOT**, the out-of-state team will need to complete enrollment **this BEFORE** appointments or tests, such as imaging, can be scheduled at an out-of-state location.
    - This goes for every individual specialist who will be seeing you or your child.
    - Why does this matter? If these individuals are not properly enrolled, claims will be denied, and the institution will not get paid for services rendered.
    - For those with commercial insurance, this may also be an issue; if this is not properly done on the front-end, insurance can deny service altogether and leave you with the entire bill to be paid as "out of network."
- Understand what your local healthcare team can provide versus what you can complete at the out-of-state clinic. All services to be completed out of state need to be documented on the referral form to obtain insurance approval on the front end.
  - o **EVERYTHING** must be approved by the state of residency's insurance before out-of-state care can be given. Most of this is done behind the scenes and families are usually unaware of this process, but it is a common reason why things get denied "last minute." Per Medicaid guidelines, many procedures and tests cannot even receive prior authorization too far in advance before a test. Thus, you cannot have something pre-authorized in April for an encounter in November. Most institutions have a policy on this; typically, they begin the submission of pre-approval paperwork 7 to 14 days before an appointment. This is why it's very important to ensure perfect accuracy in the information you provide (local provider name, insurance information, etc.)

## Tips to mitigate issues

### Medication denials

Medication denials by insurers are an unfortunate reality that can occur. It is helpful to understand why many denials are made and what can be done to prevent them. Usually, medication denials are associated with a medication that is not covered under your current drug plan, also known as a formulary or pharmacy drug list (PDL). These lists will also let you know what tier medications are and what will be required to approve your medication. If you are in a situation where your medications have been denied, do not panic. Here are some helpful steps to understand the denial.

1. Call your insurance company to clarify why the medication was denied.
  - a. Were there any coverage restrictions? One such restriction may be the need for a prior authorization. The PA approval process on average is 72 hours. Urgent PA requests may be completed within 24 hours.
  - b. Is there an emergency fill option while the denial is being approved?
  - c. Is there a quantity restriction? This is the amount (number of pills, for example) of medication prescribed. Some plans will only cover a certain amount per month.
  - d. Does this medication require step therapy?
2. Is this medication included in the formulary (or PDL)? If not, what are the formulary exceptions to this medication on your specific plan?
3. Is there an alternative to this medication that will be covered? For example, when your provider prescribes the brand name (Sabril®), but your plan will only cover generic (vigabatrin). If this substitution is allowed, then call your prescribing provider's office to let them know as soon as possible. Never assume your providing office will be notified of this denial in a timely manner.
4. Are compounded medications covered under the current plan for "off-labeled" use? Compounded medications are ones that undergo a transformation within the pharmacy to change their delivery method – for example, a medication that is typically taken in its pill form may be ground up into a fine powder and reconstituted as a drinkable liquid.

### Emergency medication needs

One way to prevent delays in medication refills is keeping track of when your medications can be refilled and starting the refill process as soon as you can, which may include needing another round of prior authorization. However, even the most precise planning may not prevent running low on medication, creating a difficult situation outside of your control. It is essential to know how to overcome this as soon as possible. Have a clear plan discussed with your prescribing provider at the time new medications are prescribed outlining what you should do in case you find yourself in the situation. Many individuals end up in this situation usually from a prolonged prior authorization process or from an unexpected denial. Rarely, lifesaving medications can experience a drug shortage. Medication drug shortages are tracked by the FDA; please reference the FDA Shortages resource in the appendix of this section for an up-to-date list.

### Medication assistance

Many medications have co-pay assistance programs for those who qualify. Generally, co-pay assistance is for commercial insurance plans. However, drug manufacturers may also have patient assistance programs (PAPs) for those who are still in need of financial assistance. While these programs often limit the total amount that an individual can utilize per year, it is helpful to know what options are available. Co-pay assistance programs are typically for medications that are brand name with no generic equivalent available. Often, your prescribing provider or specialty pharmacist can help direct how to get set up for this type of program, but they cannot do everything for you because personal financial information such as tax returns may be required.

The TSC Alliance does not provide direct financial support; however, we do advocate proactively seeking opportunities for our community to utilize to help with drug coverage with established assistance programs. Please see the Pharmacy Resource Section in the Medical Challenges section of the TSC Navigator for the most up-to-date assistance options.

Additionally, the TSC Alliance has partnered with DiRx, an online digital pharmacy platform focused on generic medicine access and affordability. This program utilizes generic formulations of medications on a cash basis only. For those who struggle with access or delays due to insurance or prior authorization, this program may offer an alternative. Programs that require Risk Evaluation and Mitigation Strategies (REMS), such as vigabatrin, are currently not available through this program. To find out more or if you have any questions, please contact DiRx customer service at 1-877-367-3479.

## Health insurance and medical benefits

Health insurance and medical benefits have various kinds of options available, including private, federal and state programs. If you are employed and have health insurance through your employer, it will most likely have a person in your human resources department to assist you with your health insurance and benefit needs and questions. If you have insurance through a federal or state program, you may or may not have a point of contact. Government services vary from one state to another. In one state you may have one person who is your contact person, whereas individuals in another state who have TSC may just have a phone number for an office that can assist them, never reaching the same person twice. Because we are covering all the issues that could occur, if you are having any issues getting on the right path, we encourage you to reach out to one of our TSC Navigators for assistance.

Be proactive on annual insurance renewals. Losing your insurance benefits can be very detrimental to yourself or your loved one. Those with Medicaid must complete forms yearly to stay active or your insurance will “lapse” or be “termed.” These are both words to indicate you no longer have an active insurance plan. Ask your insurance company if you can be assigned to a case manager (or benefits manager) who works for the insurance company to help oversee and coordinate your case, so things don’t get lost. A good case manager acts as an advocate for you because, over the years, he/she will learn which medications you have tried that have or haven’t worked. A case manager who has a good relationship with you can advocate for you when things come up like denials of payment for medications or procedures. Also, a case manager can help you to understand your co-pay and deductible requirements toward your insurance, which is vital information! Some medications for complex disorders are quite expensive and understanding your deductible requirements can make a huge impact on your health and your wallet.

Lastly, ask if your clinic has access to a social worker or case manager you can work with to help you sort out issues with insurance rejections, applying for disability if necessary or looking into other resources in the community. Be proactive annually when looking at your benefits and coverage. Make sure you reach out to the representative for your insurance plan during open enrollment to discuss any questions upfront and determine how to navigate if coverage of a specific medication or test will be changed.

Furthermore, understanding co-pays and deductible plans is essential. Remember, co-pays are forever so do not be shy if a higher deductible is cheaper over the year. There are benefits to both. For those with commercial plans, your human resources benefits department should be able to answer most questions you have.

- **Medicaid** is a federal-state assistance health care program that pays medical bills of people with low-income and limited assets. You might be eligible depending on your household income, family size, age, disability, and other factors. For more information on Medicaid, please visit the appendix of this section.
- **Supplemental Security Income (SSI)** can also be a source of monthly benefits to the disabled with low income and limited resources. Your child may qualify for SSI if he or she has a physical or mental condition that is outlined by the Social Security Administration.
- **Children’s Health Insurance Program (CHIP)** is an option for special needs children whose families are above the income level to qualify for Medicaid but too low to afford private health insurance. This program is available in all 50 states and provides coverage on prescription drugs, mental health services, vision, and hearing.
- **Waiver options:** Medicaid began as a program for low-income families. Over time, Medicaid transitioned to serving primarily children, the elderly and people with disabilities. If an adult with a disability is uninsured, requires additional services or needs wrap-around Medicaid coverage to help with finances and uncovered services, he/she may qualify for a Medicaid waiver or program. For additional information on the waivers available, specific to each state, visit [www.medicaid.gov](http://www.medicaid.gov) or your local Department of Human Services.

## Section Three Appendix

| Organization  | Description  | Website  |
|---|--|--|
| <b>Centers for Medicare and Medicaid Services (CMS)</b>       | Provides an extensive list of state-based resources on its website.  | <a href="http://www.medicare.gov/care-compare/">www.medicare.gov/care-compare/</a>                               |
| <b>Veterans Affairs</b>                                       | <i>Notable Section:<br/>Family and Caregiver Health Benefits</i>   | <a href="http://www.va.gov">www.va.gov</a>   |
| <b>Children's Health Insurance Program (CHIP)</b>             | Provides information about Medicaid and Children's Health Insurance Program (CHIP) services for families who need health insurance coverage.<br><br><i>Note: All CHIP programs are state-based, and procedures may vary.</i> | <a href="http://www.insurekidsnow.gov/">www.insurekidsnow.gov/</a>   |
| <b>Kaiser Family Foundation (KFF)</b>                         | Dedicated to filling the need for trusted information on national health issues.   | <a href="http://www.kff.org/statedata/">www.kff.org/statedata/</a>   |
| <b>The UnitedHealthcare Children's Foundation (UHCCF)</b>     | UHCCF grants help with medical expenses not covered, or not fully covered, by a family's commercial health insurance.  | <a href="http://www.uhccf.org/">www.uhccf.org/</a>   |
| <b>Medication Assistance Tool (MAT)</b>                       | MAT is a free-to-use search engine that focuses its searches on patient assistance resources available to eligible patients.   | <a href="http://www.mat.org">www.mat.org</a>   |
| <b>Needy Meds</b><br>Languages Available:<br>English, Spanish | NeedyMeds connects people to programs that will help them afford their medications and other healthcare costs.   | <a href="http://www.needymeds.org/">www.needymeds.org/</a>   |
| <b>RxAssist</b>   | RxAssist offers a database of patient assistance programs, and offers a prescription drug savings/discount card at no cost.  | <a href="http://www.rxassist.org/">www.rxassist.org/</a>   |
| <b>Step Therapy</b>   | It is a policy developed by health insurers that is intended to control costs.   | <a href="http://www.steptherapy.com">www.steptherapy.com</a>   |
| <b>Prescription Process</b>                                   | A non-profit national network of physicians who advocate for patient access to approved therapies and appropriate clinical care.   | <a href="http://www.prescriptionprocess.com">www.prescriptionprocess.com</a>                                     |
| <b>FDA Drug Shortages</b>                                     | Current and Resolved Drug Shortages and Discontinuations Reported to FDA.  | <a href="http://www.accessdata.fda.gov/scripts/drugshortages/">www.accessdata.fda.gov/scripts/drugshortages/</a> |

## Section Four: Research/Clinical Trials

### Blood and tissue donation

The TSC Biosample Repository stores samples of blood, DNA and tissues scientists can use in their research. The samples we collect are all linked to clinical data in the TSC Natural History Database.

#### Natural History Database

Implemented in 2006, the TSC Natural History Database captures clinical data to document the impact of the disease on a person's health over his or her lifetime. More than 2,500 people with TSC are enrolled in the Natural History database, and more than 900 people have contributed biosamples. The biosamples and data help researchers discover biomarkers of TSC, test potential drug treatments and determine why TSC is so different from person to person.

#### Blood samples

The TSC Biosample Repository collects samples from TSC Natural History Database participants at specific clinics and via mobile blood collections. The TSC Alliance can collect blood samples from anyone with TSC in the United States. To find out about participating and to see the most current clinical sites for the Natural History Database and Biosample Repository, please visit [www.tscalliance.org](http://www.tscalliance.org). If your current clinic is not listed, you can still be part of these opportunities by submitting the form on the website entitled "Biosample Repository Project Interest Form" or emailing [biosample@tscalliance.org](mailto:biosample@tscalliance.org).

#### Tissue samples

If an individual with TSC is having surgery to remove any part of a tissue – such as epilepsy surgery, SEGA removal or kidney or lung surgery, please contact the TSC Alliance with as much advance notice as possible at [biosample@tscalliance.org](mailto:biosample@tscalliance.org). The TSC Alliance is also able to collect cord blood and placental tissue. For post-mortem tissue donations, please see the following section.

#### Post-mortem brain donation

The University of Maryland Brain and Tissue Bank (MBTB) makes arrangements for this type of precious gift on behalf of the TSC Alliance. Individuals living anywhere in the United States who wish to donate whole brain tissue after death should contact MBTB at [btbumab@som.umaryland.edu](mailto:btbumab@som.umaryland.edu). If death is imminent and you would like to donate brain tissue, please call the MBTB as soon as possible at 1-800-847-1539. Even if not registered a staff member can be reached 24 hours a day in cases of emergencies. Successful donation rests on swift and thorough communication between family members, healthcare professionals and the MBTB. Although the MBTB will make every effort to retrieve tissue in an emergency, tissue recovery may be impossible if there is no advance notice. For more information on how to register for post-mortem donation please see the appendix of this section. There is no cost to the family to donate.

## Clinical trials and research opportunities

### What is a clinical trial?

A clinical trial (also called clinical research) is a research study using human volunteers designed to determine the safety and effectiveness of a drug, biologic (such as a vaccine), device (such as a prosthesis) or other treatment or behavioral intervention. Carefully conducted clinical trials are the fastest and safest way to find treatments that work in people and methods to improve health. Interventional trials determine whether experimental treatments or new ways of using known therapies are safe and effective under controlled environments. Observational trials address health issues in large groups of people or populations in natural settings.

### Why participate in a clinical trial?

Because clinical trials are required of any new therapy prior to FDA approval, major improvements in health care would be impossible without volunteer participants. Participants in clinical trials can play a more active role in their own health care, gain access to new investigational treatments before they are widely available and help others by contributing to medical research.

### Who can participate in a clinical trial?

All clinical trials have guidelines and criteria regarding who can participate. The factors that allow someone to participate in a clinical trial will vary from study to study. These guidelines and criteria are determined based on the goals of the study and include such factors as age, the type and stage of a disease, previous treatment history and other medical conditions. Some research studies seek participants with illnesses or conditions to be studied in the clinical trial, while others need participants without underlying health conditions. The criteria are used to identify appropriate participants needed to answer the scientific questions being asked while keeping them safe.

### What are the general benefits and risks of participating in a clinical trial?

#### **Benefits: Well-designed and well-executed clinical trials provide the best approach for eligible participants to:**

- Play an active role in their health care decisions.
- Gain access to new research treatments before they are widely available.
- Obtain expert medical care at leading health care facilities during the trial, which is generally more frequent and thorough than standard medical care.
- Help others by contributing to medical research.

#### **Risks: Clinical trials entail risks, which may include:**

- The experimental treatment may not be effective for the participant.
- The study protocol may require more time commitment than standard treatments already available. These may include increased frequency of trips to the study site, hospital stays, missed time from work and time constraints with other family dynamics.
- There may be unpleasant, serious or even life-threatening side effects to experimental treatment.

### Where do I find a clinical trial for TSC?

For a list of current clinical trials, visit the clinical trials page on the TSC Alliance website.

### Where can I get more general information about clinical trials?

The U.S. National Library of Medicine maintains [clinicaltrials.gov](http://clinicaltrials.gov), which lists research studies recruiting human volunteers in the United States and more than 200 other countries. The site also provides links to other useful resources, such as:

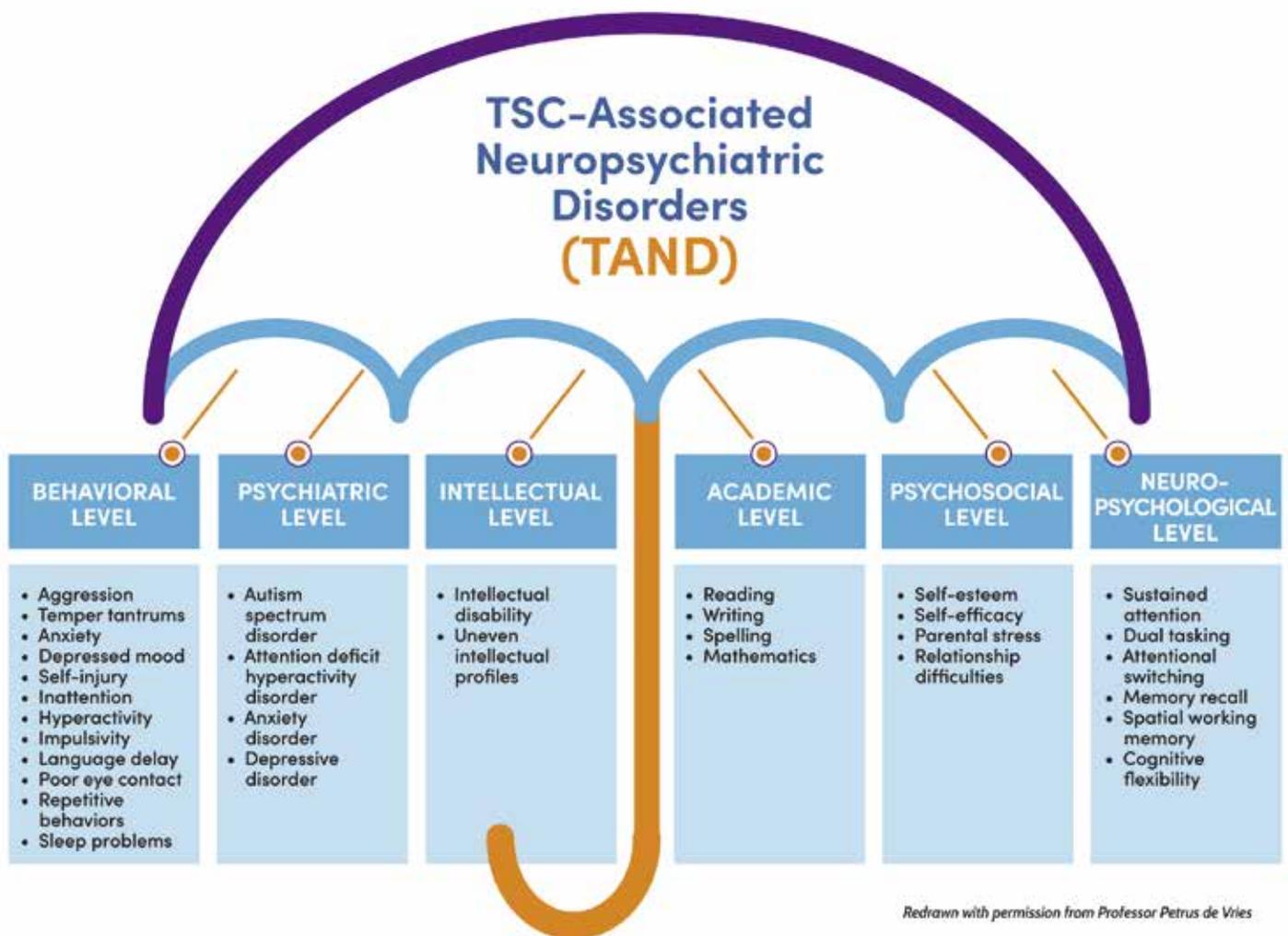
- A glossary of terms used on the [clinicaltrials.gov](http://clinicaltrials.gov) website.
- MedlinePlus®, the National Institutes of Health's website provides information about diseases, conditions, and wellness issues.
- *NIH MedlinePlus Magazine*, which presents up-to-date health information from research supported by the National Institutes of Health.

## Section Five: TSC-associated neuropsychiatric disorders (TAND)

### What is TAND?

Even though TAND affects 90% of those with TSC over their lifetime, everyone experiences unique symptoms, so interventions may be different from individual to individual. In the Behavioral Issues and TAND guide on the TSC Navigator, you will find detailed information regarding each of the impacts of the six major areas. In addition, the guide discusses behavioral intervention plans and how to put one in place as well as a functional behavior assessment. This is especially important to understand prior to student education meetings and should be shared with the school leaders.

Other helpful resources can be found on the TANDem website and behavioral management webinars by the Child Neurology Foundation under resources on the TSC Navigator. For those who are looking for therapists in your area and are unsure of who to contact you can look up your area on the National BACB therapist website.



If you have additional questions or concerns, please reach out to one of our TSC Support Navigators. If you or your loved one is in danger because of TAND-related behaviors or crises, please contact your local healthcare team or emergency services.

## Assessment of cognitive and behavioral health issues

In addition to the various physical manifestations of TSC, about 90% of TSC individuals are affected by a range of behavioral, psychiatric, intellectual, and neuropsychological complexities. This can include Autism, ADHD, intellectual disability, anxiety, and depression. TSC-Associated Neuropsychiatric Disorders, or TAND, was created to bridge the gap between identification and treatment of these complexities.

## TAND checklist

The TAND checklist was designed to help clinical teams, individuals with TSC and their families screen for TAND at every clinic visit and prioritize what the next steps are. This tool is used to screen for behavioral, psychiatric, intellectual, academic, neuropsychological, and psychosocial manifestations in TSC individuals. All individuals with TSC are at risk of having some TAND difficulties throughout their life. The severity is individual based and never the same.

## Cognitive challenges

Approximately 45% to 60% of individuals with TSC develop cognitive challenges (intellectual disabilities), although the degree of intellectual dysfunction ranges from very mild to severe. Some children appear to develop normally until the onset of seizures, when their progress slows or when they actually lose developmental milestones. Individuals whose seizures continue unchecked even after treatment (intractable seizures) have a higher likelihood of intellectual impairment. While most individuals with TSC who have intellectual disabilities also have epilepsy, many individuals with TSC who have seizures do not have significant intellectual disabilities. Some individuals with TSC may have mild learning disabilities that are essential to consider when early interventions, school programs, or career choices are being made.

## Therapies and services

TAND symptoms typically manifest from early childhood to later in life. At this phase of the journey, the main priority is to focus on early intervention and become aware of TAND symptoms so that you can have proactive discussions with your TSC providers should issues arise. The TSC Alliance has a plethora of helpful webinars and tips from experts on understanding these different manifestations and can be found on the TSC Alliance website under Video and Webinars. Additional TAND resources are provided in the appendix of this section.

## Mental health and TSC

TAND conditions can affect everyone with TSC differently. It is important to understand some of these conditions can change over the years and what might have been an issue during childhood or adolescence is now presenting much different in adulthood. In addition, those who were diagnosed with TSC later in life are often surprised that they too struggle with many conditions outlined under the TAND umbrella above. The struggle might look different, but the root cause is still the same. As with the other TSC manifestations, treatment is specific to each condition and may take trying several medications and coping strategies before finding the right combination that works best. Mental health resources continue to be an issue many in the TSC community are faced with navigating with little to no local resources.

The TSC Alliance continues to strive to improve these resources; however, we understand this does not help those of you who need resources today. If you need these resources, we strongly encourage you to discuss options your TSC clinicians can offer or refer you to locally. In addition, we recommend utilizing the National Alliance on Mental Illness (NAMI) for more tools and resources.

## Crisis intervention plan

Unfortunately, many in the TSC community will experience a crisis at some point in their lifetime. It is best to have a crisis intervention plan in place before a crisis occurs to help guide your steps when the emotions of the situation can be very difficult to process. If you or your loved one suffer with any mental health condition, it is ideal to have a plan in place that was created in a non-conflict time and can be as detailed as you feel is important. For those who know your triggers, situations or signs that usually proceed a crisis time, it is best to write those down for those in your inner circle to be aware of and can proactively intervene if possible.

NAMI provides excellent toolkits for you and or your loved one on how to prepare for a crisis as well as portable treatment records, which include what behaviors you would call 911 over or to seek emergency support. NAMI also provides a guide about navigating a mental health crisis guide that is very informative and helpful on ways to support yourself proactively or your loved one during these difficult times.

*If you or a loved one are having thoughts of self-harm or need immediate support, you can contact the National Suicide Prevention Lifeline (1-800-273-8255/TALK) or Crisis Text Line by texting "NAMI" to 741-741. In a life-threatening situation, go to your nearest psychiatric emergency room or call 911. NAMI also provides support groups and helpline for additional support and encouragement at 1-800-950-6264.*

## Documenting behaviors

Listed below are suggestions of what type of behavioral information you may want to share with your specialist. It is important to describe the observed behaviors in detail and the possible function of the behaviors, i.e., what is the trigger (reinforce) for the behavior? Describe in detail, if possible, what happened before the behaviors occurred, in what environment did the behavior occur, the duration, consequences, and outcomes from the behavior.

### EXAMPLE:

**Date/time:** 5/15/22 at 6:00 PM

**Context of the incident:** During dinner Tom started to be disruptive. His sister started laughing at him and he started screaming and throwing his food.

**Prior events before the situation occurred:** Tom had a restless sleep the night before the incident, and I took him to school in the car because we overslept. Tom and Brooke were watching television in the family room before dinner-time. Brooke laughed at Tom when he wanted to watch cartoons, then she changed the TV station and called him a baby.

- The behavior lasted for 15 minutes.
- On a scale of 1 through 5, the severity of behaviors was at a level 4 1/2.
- Tom's behavior escalated within 2 minutes.
- Stayed at a level 4 1/2 for approximately 8 minutes.
- It took about 5 minutes for Tom to return to his baseline behavior.

**Behaviors:** Tom exhibited loud screams, started throwing his food, scratching and hitting himself in the face.

**Intervention:** Tom stopped the behaviors after dad began to talk to him softly and helped him with self-feeding.

*\*Please see a sample behavior log in the appendix of this section.*

## Section Five Appendix

| Organization                               | Description   | Website  |
|--|---|--|
| <b>TANDem</b>                              | <p>TANDem is an international multi-disciplinary mobile-health project to empower families and individuals who live with Tuberous Sclerosis Complex (TSC) around the world.</p> <p><i>Notable Resources:</i><br/>           ASD and TAND<br/>           ADHD and TAND<br/>           Anxiety and TAND<br/>           Behaviors and TAND</p> | <a href="http://www.tandconsortium.org">www.tandconsortium.org</a>     |
| <b>National BACB</b>                       | Search engine for finding certified therapists in your area.  | <a href="http://www.bacb.com">www.bacb.com</a>                         |
| <b>NAMI</b>                                | <p>Dedicated to improving the lives of millions of American affected by mental illness.</p> <p><i>Notable Resource:</i><br/>           People with Disabilities</p>   | <a href="http://www.nami.org">www.nami.org</a>                         |
| <b>Ability Jobs</b>                        | Works with companies, government and nonprofit agencies to employ people with disabilities.   | <a href="http://www.abilityjobs.com">www.abilityjobs.com</a>           |
| <b>YAI Seeing Beyond Disability</b>        | YAI is a network of affiliate agencies that offer children and adults with intellectual and developmental disabilities a comprehensive range of services.   | <a href="http://www.yai.org">www.yai.org</a>                           |
| <b>Devereux Advanced Behavioral Health</b> | Devereux Advanced Behavioral Health changes lives – by unlocking and nurturing human potential for people living with emotional, behavioral, or cognitive differences.  | <a href="http://www.devereux.org">www.devereux.org</a>                 |
| <b>NeuroRestorative</b>                    | NeuroRestorative is a leading provider of subacute and post-acute rehabilitation services for people of all ages with brain, spinal cord and medically complex injuries, illnesses, and other challenges.   | <a href="http://www.neurorestorative.com">www.neurorestorative.com</a> |
| <b>Child Mind</b>                          | Child Mind is dedicated to transforming the lives of children and families struggling with mental health and learning disorders by giving them the help they need.  | <a href="http://www.childmind.org">www.childmind.org</a>               |

| Organization   | Description   | Website           |
|--|---|-------------------|
| <b>American Association of People with Disabilities (AAPD)</b> | AAPD works in coalition with other disability organizations for the full implementation and enforcement of disability nondiscrimination laws, particularly the Americans with Disabilities Act (ADA) of 1990 and the Rehabilitation Act of 1973.  | www.aapd.com      |
| <b>Centers for Disease Control and Prevention (CDC)</b>        | <p>The CDC has compiled data, research and programming specifically for people living with a disability.</p> <p><i>Notable Resource:</i><br/><i>Disability and Health Promotion</i></p>   | www.cdc.gov       |
| <b>The Mighty</b>  | <p>The Mighty is a growing community of people with lived experience sharing their honest stories. We help people connect with others around mental health, chronic illness, rare disease, disability, and more.</p> <p><i>Notable Article:</i><br/><i>Therapists with Disabilities</i></p> | www.themighty.com |



## Challenging Behaviors Log

Date: 9/15/2019 Time: 9:45 am How long did the BEHAVIOR last?: 20 minutes

Explain Incident: Logan started hitting mom and throwing objects across the room.

---

---

What happened BEFORE the behavior: Logan was asked to put his shoes on so we could go to the store.

---

---

Describe the behavior: Aggressive behavior, yelling, hitting, screaming, and throwing objects.

---

---

Severity Rating (1-5, 5 being the most severe):      1      2      3      4      5

Consequence: Logan had to pick up all of the items he threw once he calmed down. He also lost his iPad

privileges for the evening due to his destructive behavior. (This additional consequence is age appropriate for his behavior plan)

Interventions/Skills Used: Difficulty during transition period identified. Gave 5 minute warning of transition

from toys/play time to needing to put shoes on. Reinforced transition at 1 minute. Logan was removed from

his toys and sat for 2 minutes before bringing him back to the room to pick up the toys he threw.

Notes: \_\_\_\_\_

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## Section Six: Home health services

Caring for an individual with complex medical needs can be emotionally and physically exhausting for the caregiver. Depending on the needs, the countless doctor appointments, therapy services, medicines and personal care requirements may create constant worry and stress for the caregiver. Even with the best support system, this can be overwhelming. Depending on your insurance benefits, home health services may be an option. For more information about home health services, contact your local Department of Health and Human Services and request a caseworker, if you don't already have one.

### What are home health services?

Home health services are available 365 days a year, 24 hours a day to qualified individuals based on what the needs are and what insurance is willing to cover. Knowing that each case and individual has different needs, home health services can vary from qualified nurses, physical therapists, occupational therapists, speech therapists and even social workers. If your child does not need nursing care, home health services can also provide personal care services with daily activities such as bathing, toileting, eating, dressing, exercise and even playing. Home health care professionals can also assist with family outings, school activities and even respite care for the parents.

### How do I pay for these services?

Some insurance companies will help with the cost of home health services depending on the disability. State insurances, like Medicaid, may also cover up to certain number of hours/days per week or a specific monetary allotment. In addition, each state differs in hours, wages, hiring and services home health services can provide. Please note there is a difference in Medicaid coverage by state. It is best to contact your local Department of Health and Human Services for more information.

### Where do I find home health services?

The first step will be to contact your states Department of Health and Human Services and request a caseworker if you do not already have one. During this conversation make sure you are clear about your particular situation, needs and struggles. A caseworker will come to assess the environment, needs and situation. Once this is completed and approved you will be given a list of home health care agencies to contact. In some cases, states will allow you to hire someone yourself and submit for payment. In either case, please make sure to thoroughly vet the agency and/or individual to ensure they are the best fit for your loved one. Make sure to ask about accreditation, required training, background checks, screening, pediatric experience and reliability when you are researching different providers and agencies.

## Section Six Appendix

| Organization                        | Description   | Website  |
|-------------------------------------|---|--|
| <b>Special Needs Alliance (SNA)</b> | National Alliance of attorneys for special needs planning.  | <a href="http://www.specialneedsalliance.org">www.specialneedsalliance.org</a> |
| <b>Bayada</b>                       | A trusted leader in providing a full range of clinical care and support services at home for children and adults of all ages. | <a href="http://www.bayada.com">www.bayada.com</a>                             |
| <b>Pediatric Home Service</b>       | Helping kids with medical complexities  | <a href="http://www.pediatrichomeservice.com">www.pediatrichomeservice.com</a> |

## Section Seven: Legal rights under disability law

### Individuals with Disabilities Education Act (IDEA)

Infants and toddlers, birth through up to age 3 (varies by state), with disabilities and their families receive early intervention services under IDEA Part C. Children and youth ages 3 through 21 receive special education and related services under IDEA Part B.

Once a child turns age 3, IDEA requires public school systems to develop appropriate Individualized Education Programs (IEPs) for each child from age 3 to the age of majority in each state. The specific special education and related services outlined in the IEP reflect the individualized needs of each student. Each student's IEP must be developed by a team of knowledgeable persons (which includes the child's teacher, parents/caregiver, special education representative and other individuals as requested by the parent or other agencies involved). The IEP must be reviewed at least annually.

For more information on the IDEA 2004 visit U.S Department of Education.

### Section 504

The term "504" comes from the Rehabilitation Act of 1973 (Section 504), which is a civil rights law that states a child with a disability is entitled to a free, appropriate public education the same as a child without a disability. This law ensures children with disabilities do not face barriers to receiving an education. Unlike the Individuals with Disability Education Act (IDEA), which is an education law that mandates a child receive the necessary educational supports and services to progress in the general education curriculum, the intent of 504 is to prevent discrimination in not providing equal access to education. Section 504 covers qualified students with disabilities who attend schools receiving Federal financial assistance. To be protected under Section 504, a student must be determined to: (1) have a physical or mental impairment that substantially limits one or more major life activities; or (2) have a record of such an impairment; or (3) be regarded as having such an impairment. Section 504 requires that school districts provide a free appropriate public education (FAPE) to qualified students in their jurisdictions who have a physical or mental impairment that substantially limits one or more major life activities. Section 504 is more about accommodations than special education services.

Some individuals with TSC may not be eligible for special education and support services but still require accommodations so they can participate in school activities. Please visit the TSC Alliance website for more information on school issues.

Under Section 504 if the child's disability affects his or her access to learning, he/she is entitled to accommodations under 504. For more information about Section 504 please see the resources listed at the end of this section.

If you need help with navigating the education system, please reach out to the TSC Alliance for support by completing the IEP Intake Form located under School Issues on the TSC Alliance website.

### Legal assistance

The TSC Alliance acknowledges that legal assistance is sometimes needed to assist with certain barriers or challenges that are unique to your situation for appropriate resolution. However, we cannot endorse an individual or organization.

### Family and Medical Leave Act (FMLA)

There are times when a parent or caregiver will have to take extra time off work. It may be for a series of specialist appointments, testing, procedures or hospitalizations. The Family and Medical Leave Act, known as FMLA, allows covered employees to take up to 12 weeks (480 hours) of UNPAID leave if they have a serious health condition or need to care for an immediate family member (parent, son, daughter, spouse) with a serious health condition. FMLA is a job-protected leave for employees. Check with your employer about FMLA and contact HR for the required paperwork. For more information about FMLA please visit the U.S Department of Labor.

## Air Carrier Access Act

The Air Carrier Access Act prohibits discrimination based on disability in air travel. The Department of Transportation has a rule defining the rights of passengers and the obligations of airlines under this law. This rule applies to all flights of U.S. airlines, and to flights to or from the United States by foreign airlines. The COVID-19 pandemic has created provisions for travelers regarding the mandatory safety requirements to those with disabilities. Given the ongoing and unforeseen changes to this pandemic, we recommend visiting the U.S Department of Transportation: Passengers with Disabilities for more information. In addition, you can reach out to the airlines directly regarding current safety requirements or concerns.

## Telecommunications Act

The Federal Communications Commission (FCC) rules under Section 255 of the Communications Act require telecommunications equipment manufacturers and service providers to make their products and services accessible to people with disabilities if such access is readily achievable. Where access is not readily achievable, manufacturers and service providers must make their devices and services compatible with peripheral devices and specialized customer premises equipment that are commonly used by people with disabilities if such compatibility is readily achievable.

## Section Seven Appendix

| Organization  | Description  | Website  |
|---|--|--|
| <b>Social Security Administration</b>   | Provides information about eligibility and application requirements. The SSA website provides answers to frequently asked questions, forms, and online tools to help determine eligibility for certain benefit programs. | <a href="http://www.ssa.gov/disability/">www.ssa.gov/disability/</a> |
| <b>U.S. Department of Justice - Civil Rights Division</b>                         | A guide to disability rights laws  | <a href="http://www.ada.gov/cguide.htm">www.ada.gov/cguide.htm</a>   |
| <b>Learning Disabilities Association of America (LDA)</b>                         | LDA's mission is to create opportunities for success for all individuals affected by learning disabilities through support, education, and advocacy.   | <a href="http://www.lidaamerica.org">www.lidaamerica.org</a>         |
| <b>The Council of Parent Attorneys and Advocates (COPAA)</b>                      | An organization of attorneys, advocates and parents established to improve the quality of legal assistance for parents of children with disabilities.  | <a href="http://www.copaa.org">www.copaa.org</a>                     |
| <b>US Department of Labor (DOL) Office of Disability Employment Policy (ODEP)</b> | Provides fact sheets regarding disability issues, discrimination, and legal rights   | <a href="http://www.dol.gov/odep">www.dol.gov/odep</a>               |

## Section Eight: Educational rights

### Services under IDEA

As discussed in Section Four, the Individuals with Disabilities Education Act (IDEA) is a law that makes available a free appropriate public education to eligible children with disabilities throughout the nation and ensures special education and related services to those children. The IDEA governs how states and public agencies provide early intervention, special education, and related services to eligible infants, toddlers, children and youth with disabilities. Navigating the educational journey with TSC can be overwhelming but there are resources and support available to assist in the process through the TSC Alliance.

Special education is instruction that is specifically designed to meet the unique needs of children who have disabilities and is provided at no cost to parents. This can include special instruction in the classroom, home, hospitals or other settings. The TSC diagnosis alone does not mean a child will automatically qualify to receive special education services in school. Under the law, the Individuals with Disabilities Education Act of 2004 requires public schools to provide special education services to children ages 3 to 21 who meet certain criteria. To qualify, a student must:

- Have a documented disability in one of the 13 documented categories covered by IDEA (you can find the categories in the appendix of this section), and
- Need special education in order to access the general education curriculum.

If your child is not currently receiving early intervention or special education services, and you think these services are needed, the first step is to find out if your child has a disability as defined by the IDEA by requesting a special education evaluation.

### School evaluations

To initiate the process, you will need to request in writing a special education evaluation. The letter should include the teacher, the principal, and the special education director of the school. Let them know you believe your child has a disability and might need special education support. Ask the school to evaluate your child as soon as possible. You can view a sample template letter in the appendix of this section to request an evaluation. The school may also approach you about the need for a special education evaluation.

The school may not think your child has a disability that requires special education services and can refuse a request for a special education evaluation. The school must provide the decision in writing, as well as a cogent reason for the refused evaluation. If the school refuses to evaluate your child, there are two things you can do immediately:

- Ask the school for the board approved special education policies and your procedural safeguards (the rights and procedures for parents who disagree with decisions made by the school system). These materials outline the steps to take for dispute resolution options for parents/caregivers when they disagree with the school decision. They include requesting a meeting with the school personnel, or more formal dispute resolution options like state complaint, mediation or due process, which may require an attorney. If you need support, please contact the TSC Alliance via IEP Intake Form on our website. If you do not have access to a computer, contact Shelly Meitzler at [smeitzler@tscalliance.org](mailto:smeitzler@tscalliance.org) or 1-800-225-6872.
- You can privately pay for an Independent Education Evaluation (IEE) to demonstrate the need for special education services. If you choose to pay for an IEE make sure the evaluator knows that the evaluation must meet the school district's criteria. The school is not required to implement the recommendations or findings of the IEE, but it must consider the data in deciding if your child needs services.

When the school approves the request to evaluate before anything can begin, it will need written consent. The school will provide a Permission to Evaluate form that must be signed and returned. It is good practice to make copies of all documents, making sure you have accurate up-to-date records and ensure timelines and next steps are being carried out by whomever is responsible. The evaluation process can take up to 60 days and varies by state based on regulations.

To get special education services a child must be evaluated and determined eligible in one of the 13 qualifying categories. The following procedures must be adhered to in evaluating and qualifying a child with disability under IDEA 2004.

All testing or evaluations must:

- Not discriminate based on race or culture
- Be given in the child's native language or mode of communication unless it is clearly not feasible to do so
- Use a variety of tools and strategies
- Be validated for the purpose used
- Be given by person trained/knowledgeable
- Be used in accordance with the test instructions
- Measure more than just IQ
- Accurately measure aptitude and/or achievement
- Use numerous tests in the evaluation process

If a child has more than one disability, he/she will qualify under the disability that most adversely affects learning, or he/she can qualify under multiple disabilities. If your child has intellectual disabilities and autism, he/she could be identified under either depending on which is affecting learning the most. If one does not outweigh the other, then you might want your child evaluated for multiple disabilities.

The evaluation team may include the school psychologist doing the actual testing, as well as others working as a team throughout your child's evaluation process. They give the child various tests and review school records and obtain parent input. They also observe in the classroom. This team might include a classroom teacher and a special education teacher. One important player on the team is the parent/caregiver.

If the evaluation shows your child has a disability, the next step is for the school to determine whether he or she needs special education services. Once the evaluations are completed and the reports are submitted with the findings, an eligibility meeting will be held to review the results and to determine if the student is eligible for special education services.

You should be provided with a copy of the evaluation reports prior to the meeting so you have sufficient time to read and understand the results. You have the right to ask questions and provide any input during the meeting as well. Always follow up after any meeting summarizing your understanding of the meeting and any notes you want included.

If the school determines your child needs services, the next step is to create an Individualized Education Program (IEP).

## Finding the right classroom placement for your child to be successful

The Federal law Individuals with Disabilities Education Act outlines the Least Restrictive Environment (LRE). The LRE means to the maximum extent possible and appropriate, children with disabilities should be educated with children without disabilities in the general education setting. Only after all appropriate aids, services and other supports identified for the student have been exhausted should removal from the general education setting be considered. This decision is made annually, including all members of the IEP team and based off the appropriate needs identified in the IEP. Appropriate means the education meets the student's special needs and allows the student to make educational progress.

- **Inclusion Classroom** – In this placement, your child will be in a regular education class with peers their age. In addition to the regular education teacher, a special education teacher will adjust the curriculum to your child's specific educational needs.
- **Resource Room** – This placement provides more intensive help to stay at grade level. The student will work with a special education teacher in a small group but will stay in the regular education setting.
- **Self-Contained Classroom** – Students in a self-contained classroom are working at all different educational levels. Some will have regular education time built into their schedules when they will go to a regular education classroom for part of the day, sometimes with an aide. The student-to-teacher ratio is smaller in a self-contained classroom. Self-contained classrooms offer routine, structure, and individual appropriate expectations.
- **Out-of-District Placement** – Sometimes when your home district cannot meet the needs of the child, as identified in the IEP, an out-of-district placement may be needed. These placements are often used to address special learning and behavioral needs.

It is hard to decide which classroom placement is appropriate for your child. We all want what is best for our children and this is no different. Just know that a classroom placement is never permanent.

## Developing an Individual Education Program (IEP)

If a child is found eligible for special education services, you and the school will collaborate to design an Individual Education Program (IEP). An IEP must be developed within 30 calendar days from the date that a child is determined to be eligible for special education services. The parent must sign the initial IEP.

### Preparing for an IEP meeting

There are several steps parents can take to prepare themselves for their child's first IEP meeting. Taking these steps prior to the meeting may assist you in advocating for the services your child needs.

Review all documentation prior to the meeting. This may include the current draft IEP, recent progress reports and report cards. Collect samples of your child's homework, tests, and notes from the teacher. Gather your own notes and observations and any other relevant information including outside evaluations, doctor notes or recommendations.

- **Be objective:** Use objective measurements that illustrate your child's struggles. For example, "it takes my child 3 hours to complete a reading assignment, whereas most children his age finish it in a half hour." The more data available helps validate the need for a service or support.
- **Bring support:** Bring your spouse, significant other, a friend, or family member who can provide support and/or help take notes during the meeting. It can be an emotional meeting for parents/caregivers. The TSC Alliance has support available if you would like someone on our staff to attend a school meeting.
- **Take good notes:** Keep track of the discussion that takes place during the meeting. Make sure to note any action items, the person they were assigned to and any follow up actions to summarize in the written follow up for clear communication. Be sure to make all requests in writing. Below is an example.

| Grade | IEP/504 Date | Evaluation Date | Concerns  |
|-------|--------------|-----------------|---|
| K     | 10/15/21     | 10/4/21         | Logan is having a hard time with staying on task, he is easily distracted. Logan has difficulties with recalling letters and numbers when he was able to in the past. |

Members of an IEP Team should include the following:

- Parents/guardians of the child
- Regular education teacher of the child if the child is or may be in the regular education environment
- Special education teacher of the child or where appropriate, not less than one special education provider for the child
- District representative
- A person to interpret evaluation results
- Child, when appropriate
- Other individuals with specific knowledge or expertise in the area of the child's disability

Prior to IDEA 2004, all these participants were required to attend the IEP meeting for the IEP to be written. Now, a member of the IEP team can be excused from attending an IEP meeting if the IEP team member's area is not being modified or discussed, and if the parent and school agree in writing the member's attendance is not necessary.

If the school system wants to excuse an IEP team member when his/her area is being modified and discussed then the law states that a parent must be fully informed in his or her native language, or other mode of communication, and understand the granting of consent is voluntary and may be revoked at any time. The excused member must also provide in writing to the parent and the IEP team input into the development of the IEP prior to the meeting.

The IEP team member who is the school district representative must be qualified to provide, or supervise the provision of, specially designed instruction, and be knowledgeable about the general education curriculum and the availability of resources of the school. They must also have the authority to commit those resources. This means that if you are in an IEP meeting and the team has decided your child needs occupational therapy (OT) for 20 minutes twice a week, the district representative has the ability to state the school will provide those services. If you are in a meeting and the district representative states he or she does not have the authority, then all your required team members are not present, and the meeting will need to be rescheduled until a district representative is available who can commit to services.

### **Prior Written Notice**

You should receive a written notice requesting that you attend your child's IEP meeting. This notice should include the proposed date, time and location. It should also include alternate dates and times to accommodate your schedule. The law states IEP meetings should be held at a time and place convenient to parents.

### **Understanding IEP components**

An IEP is a written statement of the educational program designed to meet a child's individual needs. Every child who receives special education services must have an IEP. Once the IEP is developed, services should be implemented as soon as possible following the initial IEP meeting. It is a working document and is used throughout the school year to monitor educational progress. Your child's school team should have access to your child's IEP and understand the responsibilities in accommodations, modifications, supports and services that will be provided.

### **IEP requirements**

The IEP must include the following information:

- Statement of the student's present levels of academic achievement and functional performance (PLAAF).
- Statement of measurable annual goals, including short-term objectives for some students.
- A description of how the child's progress toward meeting the annual goals will be measured and when progress reports will be provided.
- Statement of special education and related services, supplementary aides, supports and services, modifications/accommodations, and personnel.
- Explanation of the extent the student will not participate in regular education and/or extracurricular and non-academic activities.
- Statement of any individual appropriate accommodations necessary to participate in state and district-wide achievement tests.
- IEP starting dates ending dates, frequency, duration and location of all services.

### **Requesting IEP amendments**

An IEP is reviewed annually; however, this doesn't mean a meeting can't be requested at any time. Examples of additional meetings can include changes in medical history such as brain surgery, review of annual goals based off progress reports and any concerns that impact the supports and services identified in the IEP.

### **Present Levels of Academic Achievement and Functional Performance (PLAAF)**

The Present Levels of Academic Achievement and Functional Performance (can be referred to as PLAAF, PLEP, PLOP or PLP) describes the child's current abilities, skills, weaknesses and strengths — academically, socially, and physically. To gather this information, the IEP team must look at the whole child: the child in the regular education classroom, the child interacting with their peers, how the child performs at home, etc. How does the child perform next to their peers the same age? How does a child's disability interfere with progress in the general education curriculum?

### **Measurable annual goals**

When a goal is included in the IEP, there must be a present level of academic achievement and functional performance, or baseline, established. When the IEP team writes an annual goal, everyone must agree to the projected progress. The team should write goals that are SMART: specific, measurable, attainable, results oriented and time bound.

The goal should be stated in a way that progress can be measured by standardized tests, curriculum-based measurement or screening. Your role as a parent with meaningful parental participation is to ensure you understand all parts of the IEP. Ask questions and do not hesitate to ask for additional information or clarification if you do not understand something. If the child is of transition age, make sure the goals include transition planning.

**Example:** *John will be able to read CVC words with 80% accuracy by the end of the school year. Do you know what a "CVC word" is? Instead of writing "CVC words," the IEP should read: "John will be able to read Consonant-Vowel-Consonant words like cat, rat, and sat, 8 out of 10 tries by the end of the school year."*

## Medical issues

A Seizure Action Plan should be included in an IEP to address all seizure protocol. If there are additional health concerns, an Individualized Health Plan (IHP) can be developed and included as part of the IEP.

## Seizure Action Plan

For all individuals with seizures, it is important to provide the school with a Seizure Action Plan (SAP). A SAP helps organize all the individual's seizure information to guide the school on what to do in an emergency. There are some great resources available from the Seizure Action Plan Coalition and the Epilepsy Foundation. Please follow the link in the resources at the end of this section for more information. In addition, those within your inner circle (friend, family, school staff) can complete additional training on seizure safety for an extra layer of confidence to help ensure your loved one receives high quality seizure support in the event it is needed.

## Additional services in the school setting

Special education includes a description of the type of support your child will receive and in what setting. The law states that children are to be educated in the general education setting to the maximum extent possible. Only if a disability is so severe that supplementary aids and services cannot provide the child with an appropriate education should he/she be placed in a more restrictive environment. This will be identified by how much time will be spent in each appropriate setting.

Related services help children with disabilities benefit from their special education by providing extra help and support in needed areas to benefit from general education. There is an extensive list, but some examples based on individualized need can include: speech, physical or occupational therapy, transportation, nursing services, interpreter services and parent counseling and training.

This section will identify all specially designed instruction, related services and/or supplementary supports/services and will clearly define who will provide the service, where and how often the service will be delivered. It will also include accommodation and modifications.

| Therapy | How Often        | Who              | Where                     |
|---------|------------------|------------------|---------------------------|
|         | 2x weekly 20 min | Speech Therapist | Small Group/Resource Room |

## IEP guidance

The TSC Alliance recognizes parents and young adults often face many difficulties regarding educational and school-related issues.

The TSC Alliance is proud to manage an incredible group of volunteers who provide local resources in most US states and staff who can provide guidance on education issues and training for local school districts on how learning is unique for people with TSC. If you would like help navigating educational resources for someone with TSC, please begin by completing the IEP intake form. This IEP intake form provides the necessary information about the school system and support needed with the education process to the TSC Alliance staff. You can access the IEP intake form on the TSC Navigator under School Issues.

## Homeschooling

Homeschooling is regulated by state law and is allowed in all 50 states (as well as all U.S. territories). Each state has its own legal rules and requirements.

It is strongly encouraged that you research the regulations and requirements specifically if your child has been identified under the Individuals with Disabilities Education Act (IDEA) as needing special education. For more information, visit Home School Legal Defense Association (HSLDA) and your state department of education.

## Section Eight Appendix

| Organization  | Description  | Website  |
|---|--|--|
| <b>Department of Education</b>  | Our mission is to promote student achievement and preparation for global competitiveness by fostering educational excellence and ensuring equal access.  | <a href="http://www.ed.gov">www.ed.gov</a>   |
| <b>Individuals with Disabilities Education Act (IDEA)</b>                                       | Access from the U.S Department of Education website  | <a href="http://www.sites.ed.gov/idea/about-idea/">www.sites.ed.gov/idea/about-idea/</a>                             |
| <b>Office of Civil Rights (OCR)</b>   | Access from the U.S Department of Education website  | <a href="http://www.ed.gov/about/offices/list/ocr/aboutocr.html">www.ed.gov/about/offices/list/ocr/aboutocr.html</a> |
| <b>Home School Legal</b>  | <p>HSLDA advocates for the freedom to homeschool and offers support for every stage of the homeschool journey.</p> <p><i>Notable Resource:</i><br/><i>Homeschool Laws by State</i></p>   | <a href="http://www.hslda.org">www.hslda.org</a>   |
| <b>Council for Exceptional Children</b>   | The Council for Exceptional Children (CEC) is the largest international professional organization dedicated to improving the success of children and youth with disabilities and/or gifts and talents. CEC advocates for appropriate governmental policies, sets professional standards, provides professional development, and helps professionals obtain conditions and resources necessary for effective professional practice.   | <a href="http://www.exceptionalchildren.org">www.exceptionalchildren.org</a>   |
| <b>Center for Parent Information &amp; Resources</b><br><i>Available in English and Spanish</i> | <p><i>Notable Articles:</i><br/> <i>"Developing Your Child's IEP"</i><br/> <i>"When the IEP Team Meets"</i><br/> <i>"Questions Often Asked by Parents about Special Education Services"</i><br/> <i>"Contents of the IEP"</i><br/> <i>"Special Factors in IEP Development"</i></p>   | <a href="http://www.parentcenterhub.org">www.parentcenterhub.org</a>   |
| <b>Understood for All Inc.</b><br><i>Available in English and Spanish</i>                       | <p>Sample letter requesting evaluations and reports for services.</p> <p><i>Notable Articles:</i><br/> <i>"What is Early Intervention?"</i><br/> <i>"The difference between IEPs and 504 plan"</i><br/> <i>"What is Special Education?"</i><br/> <i>"4 Benefits of Inclusive Classrooms"</i><br/> <i>"What is the Individuals with Disabilities Education Act (IDEA)?"</i><br/> <i>"The 13 disability categories under IDEA"</i></p> | <a href="http://www.understood.org">www.understood.org</a>   |

| Organization               | Description  | Website  |
|----------------------------|--|--|
| <b>Seizure Action Plan</b> | <p>This collaborative effort is designed to raise awareness of what a Seizure Action Plan (SAP) is the importance of a SAP in the health management of those with epilepsy; and resources to develop an individualized SAP.</p> <p><i>Notable Resource:</i><br/> <i>School Staff: how to respond to a seizure Seizure Action Plan</i><br/> <i>First Aid Training for School Nurses and Personnel</i></p> | <a href="http://www.seizureactionplans.org">www.seizureactionplans.org</a> |



## IEP Meeting Information:

Date: 3/14/21

Teacher: \_\_\_\_\_ Email: \_\_\_\_\_

School Representative: \_\_\_\_\_ Email: \_\_\_\_\_

Physical Therapist: \_\_\_\_\_ Email: \_\_\_\_\_

Occupational Therapist: \_\_\_\_\_ Email: \_\_\_\_\_

Speech Therapist: \_\_\_\_\_ Email: \_\_\_\_\_

School Nurse: \_\_\_\_\_ Email: \_\_\_\_\_

Other: \_\_\_\_\_ Email: \_\_\_\_\_

## Notes:

Based on everyone's assessment, Logan is reaching his goals and they are able to amend adding more challenging goals to work towards. Logan enjoyed going to Mr. Spegals class for GenED time. He goes for calendar, science, specials, math centers and reading centers. Mrs. Fracassa agrees that Logan would benefit from a 1:1 aide as he needs constant redirection and assistance to complete tasks. Also would help to monitor seizures.

Goals Reached: Logan is able to count by 1s, 5s, and 10s to 100. He is able to add and subtract up to 15, up to 20 with assistance. Logan has mastered CVC words in reading.

Future Goals: Reading: reading using picture clues, Math: add/subtract up to 20 WITHOUT assistance (extend to 25), OT: continue to work on fine motor skills and handwriting, SLP: continue to work of conversational speaking (answering questions and following up with his own question)

## Section Nine: Transition planning

Regardless of their disabilities, all children eventually become adults – both physically and legally. Transition planning is an intentional and coordinated process used to help students with disabilities transition from school to post-school life. Per IDEA, planning can begin as early as 14 years old and varies by state but must be implemented by 16 years old.

The transition plan is a part of the IEP and is a collaboration between the parent or guardian, student (when appropriate), special education teacher, regular education teacher, administrative representative (in some cases a school transition coordinator), vocational rehabilitation, counselor and/or someone from adult day services who may be part of the transition team.

Discuss your child's graduation plan with the IEP team to ensure it is appropriate for your child. The IEP team is responsible for determining whether a student will need to meet the state's graduation requirements or a modified set of requirements. If a student is receiving a modified curriculum (rather than just accommodations that provide access to the same coursework as their general education peers), it is possible he or she is on a "certificate track," which means the student will receive a certificate of attendance/completion but is not earning credits to graduate with a diploma.

It will include an age-appropriate transition assessment (AATA). AATA is an assessment of the student's interests, needs, preferences and strengths assessment to determine the Present Level of Academic Achievement and Functional Performance (PLAAF) statement. It should include formal and informal assessments. The data collected from both the PLAAF and the AATA will help write the post-secondary goals for the individual.

Some questions to consider when planning:

What activities does the child enjoy?

- What types of chores can they complete around the house?
- Who will they live with?
- How will the child travel around?
- Will the child need a guardian?
- What financial support will be needed?

### Transition goals

Transition goals are a part of the IEP and are measurable and written with a result in mind. The goals should be specific, measurable, attainable, relevant and time bound.

The goals should identify the appropriate level of support needed to assist with post-secondary options, which can include college or a vocational/trade school; finding employment, which can include earning a living, internships, supported employment, full-time job or volunteering; and community living, which could include independent living with functional life skills such as budgeting, self-care, mobility and transportation as well as community experiences.

Transition support can include specially designed instruction, courses of study, related services, employment, daily living skills and community experiences.

### Health care transition (HCT)

Transitioning from pediatric to adult health care is something all children will experience regardless of if they stay with pediatric TSC experts or if they transition to adult care providers. Either way, the healthcare sector changes as well as healthcare insurance and the liability and scope of practice of many pediatric providers. Having a solid understanding of what this transition looks like and what you can do to prepare yourself and your child is the best approach to ensuring high quality care across the lifespan.

The health care transition plan should be initiated by your child's medical team, however, as the entire pediatric community continues to make this a priority. Depending on where you are on your journey, this might be patient/family driven. The following information will focus on when this transition should be started and important factors to consider from a medical aspect along the way.

Health care transition (HCT) should begin around the ages of 12 – 14. During this period start having those discussions with your medical team expectations, concerns and barriers that will impact your child’s medical team. Depending on your child’s functional level, a transition will look different for those who are independent vs. those who will be dependent adults. Learning the adult health care system still applies if your TSC medical team will not change. As your child becomes an adult, it is important to establish a primary healthcare provider. Internal medicine or comprehensive/complex care management providers (dually certified in pediatrics and internal medicine, known as “Med-Peds certified”) are two great places to start as you transition from a pediatrician to adult focused care.

Ages 14 – 18 are key times to track life skills, self-care skills and educational needs surrounding medical care and awareness to gauge readiness for transition. This can be as simple as your child being able to pronounce what medications he/she takes and why, the frequency of the medication and the dosage of the medications all the way to understanding the manifestations of TSC and what each role the medical care team contributes to overall health and well-being. A critical piece is to start searching for those additional adult providers who will become more frequent in your child’s healthcare journey. It is important to initiate an encounter (visit) during this age and can continue to be included in subsequent visits until the hand off of clinical care has shifted.

We are currently in the process of working with our recognized TSC Clinics and TSC Centers of Excellence to establish seamless transition plans and guidance to assist with this process. Please continue to check the TSC Navigator for these updates. For those of you who are closer to the transition, you can find additional information on the TSC Navigator under Resources and then Caregiver Resources for more specific information around the adult health care model and things to be aware of as you the caregiver ages too.

### **Transition talking points**

- Make sure those with intellectual disabilities have formal neuropsychological testing
- Discuss options such as conservatorship vs. guardianship
- Traditional schooling (college) or job placement options
- Estate and financial planning
- Seizure Action Plans, rescue meds and SUDEP
- Educating about what to expect during the timelines, understanding the key differences between pediatric and adult care models and how to advocate for unique needs

## Section Nine Appendix

| Organization  | Description  | Website  |
|---|--|--|
| <b>The Understood</b>   | <i>Notable Resource:<br/>IEP Transition Planning<br/>Sample IEP Transition Plan and Goals</i>  | <a href="http://www.understood.org">www.understood.org</a>                             |
| <b>PACER</b>  | <i>Notable Resource:<br/>Transition IEP Checklist</i>  | <a href="http://www.pacer.org">www.pacer.org</a>                                       |
| <b>Department of Education</b>                                    | <i>Notable Resource:<br/>A Transition Guide: to postsecondary education and employment for students and youth with disabilities</i>  | <a href="http://www.ed.gov">www.ed.gov</a>   |
| <b>National Technical Assistance Center on Transition (NTACT)</b> | Is a Technical Assistance Center co-funded by the U.S. Department of Education's Office of Special Education Programs (OSEP) and the Rehabilitation Services Administration (RSA) which provides information, tools, and supports to assist multiple stakeholders in delivering effective services and instruction for secondary students and out of school youth with disabilities.<br><br><i>Notable Resources:<br/>Postsecondary Education<br/>VR Transition Services<br/>Pre-Employment Transition Services<br/>Instruction in Self-Advocacy</i> | <a href="http://www.transitionta.org">www.transitionta.org</a>                         |
| <b>Got Transition</b>   | Got Transition aims to help youth and young adults move from pediatric to adult health care.   | <a href="http://www.gottransition.org">www.gottransition.org</a>                       |
| <b>Child Neurology</b>  | <i>Notable Resource:<br/>Transition of Care Toolkit</i>  | <a href="http://www.childneurologyfoundation.org">www.childneurologyfoundation.org</a> |

## Section Ten: Employment, vocation and housing

### Employment rights

The Americans with Disabilities Act (ADA) is a federal mandate that prohibits discrimination based on disability in the areas of employment, housing, public accommodation, transportation and telecommunication services. Title I of the ADA states employment practices cannot discriminate against a person with a disability who is qualified for the job. A person with a disability who can complete the essential functions of the job is considered qualified. The law also requires an employer to provide “reasonable accommodations” or modifications to a work environment to guarantee people can effectively perform their jobs, such as providing wheelchair accessibility. For more information, please visit [www.eeoc.gov](http://www.eeoc.gov).

### Vocational/rehabilitation programs, jobs and employment

For some people, participating in a vocational, educational or other type of activity can be empowering and may provide purpose. Vocational and rehabilitation options for people with disabilities range from positions that provide high levels of assistance to competitive employment without additional accommodations. Experience provided through work, job training or other enrichment activities may add to the quality of life and increase a person's self esteem. Additionally, social interactions with coworkers may enhance a person's integration in the community.

Often the challenge is not in performing the job, but in finding the job that best suits an individual's interests, strengths and personality. One of the best places to begin a job search is with your state vocational rehabilitation (VR) agency. State VR agencies coordinate and provide several services for people with disabilities looking for a job. These services may include counseling, skills evaluation, training, job placement, coaching and support. Services provided vary by state, but most agencies will assign a vocational counselor to work with an individual with a disability to identify and locate employment options that best suit their capabilities, needs and interests. State VR agencies work with non-profit organizations and private employers that may provide a wide range of employment options in a variety of settings. You can find your state or local VR agency by checking your state government websites or by visiting the Job Accommodation Network.

### Federal employment and support programs

**Ticket to Work** is a nationwide initiative that offers Social Security Administration (SSA) disability beneficiaries greater choice in obtaining services they need to help them go to work. To qualify, you must be a SSA disability beneficiary. This program may be able to assist you in finding employment, vocational rehabilitation and other support services from public and private providers. For more information about Ticket to Work, visit [choosework.ssa.gov](http://choosework.ssa.gov).

**Project Employ** is a joint initiative of the Office of Disability Employment Policy and the Society for Human Resource Management in partnership with other employers to promote and expand employment opportunities for people with significant cognitive disabilities such as intellectual disability, autism, psychiatric disorders and autism. Project Employ works with groups such as Goodwill Industries International, Association for Persons in Supported Employment, Administration on Developmental Disabilities and others. Employer (private sector) partners include companies such as DuPont Pharmaceuticals, AT&T, Marriott Foundation, General Electric, IBM, Southwest Airlines and Prudential Insurance Company of America. Programs vary by state; however, Project Employ provides a variety of career development skills and training to people with disabilities, which are provided through a network of employment training and placement providers that specialize in serving people with severe disabilities.

**The Job Accommodation Network (JAN)** is a free consulting service of the U.S. Department of Labor Office of Disability Employment Policy that provides information about job accommodations, the Americans with Disabilities Act and the employability of people with disabilities. JAN is not a job placement service; however, it does provide services that assist people with disabilities become informed about their educational rights, acquire accommodation options and learn about other government and placement agencies.

**Association of University Centers on Disabilities (AUCD)** hosts a nationwide network of interdisciplinary centers advancing policy and practice for and with individuals with developmental and other disabilities, their families and communities. At least one center per state provides a wide range of community outreach and assistance in the areas of special education, employment and housing programs. All centers are part of universities or medical centers and serve as a link between the university and community. Services provided by each center will vary. For example, some provide more services to adults with disabilities. In some instances, centers may work with the local Developmental Disability Planning Council in providing services and information.

**Developmental Disability (DD)** Planning Councils are assisted by the U.S. Department of Health and Human Services (HHS) and provide information on disability-related organizations in your state. The information offered by each council differs by state. To find the DD council in your state, visit [www.nacdd.org](http://www.nacdd.org).

## How do I find a job?

One of the best places to begin a job search is with your state vocational rehabilitation agency. State vocational and rehabilitation (VR) agencies coordinate and provide several services for people with disabilities looking for a job. These services may include counseling, skills evaluation, training, job placement, coaching and support. Services provided vary by state, but most agencies will assign a vocational counselor to work with an individual with a disability to identify and locate employment options that best suit their capabilities, needs and interests. The state VR agency will also be able to help individuals determine the services and programs for which they qualify. State VR agencies work with nonprofit organizations and private employers that may provide a wide range of employment options in a variety of settings. For a link to the vocational rehabilitation agency in your state, visit [www.ed.gov](http://www.ed.gov).

## Housing and independent living

### Residential programs

During the transition phase and estate planning, individuals and families should consider housing options that best serve the needs of the adult with TSC. These needs will vary between each individual and family. It is important to research the options available in your state before the time they are needed. While all states receive federal funding to provide these services, options vary from state to state, and many have long wait lists. There are many things to consider when thinking about housing options such as independent living, independent living community, semi-independent and residential housing communities that may include 24-hour care facilities including nursing homes and long-term living facilities. For those who will live at home, other options are available such as skill development and adult day programs.

### Public housing

Through the Section 811 Supportive Housing for Persons with Disabilities program, the U.S. Department of Housing and Urban Development (HUD) provides funding to develop and subsidize rental housing with the availability of supportive services for very low- and extremely low-income adults with disabilities. For more information about HUD, please visit the appendix of this section.

### Section 811

The newly reformed Section 811 program is authorized to operate in two ways: (1) the traditional way, by providing interest-free capital advances and operating subsidies to nonprofit developers of affordable housing for persons with disabilities; and (2) providing project rental assistance to state housing agencies.

## Section Ten Appendix

| Organization  | Description  | Website             |
|---|--|---------------------|
| <b>Job Accommodation Network (JAN)</b>  | A source for free, expert, and confidential guidance on job accommodations and disability employment matters.  | www.askjan.org      |
| <b>HUD U.S Department of Housing and Urban Development</b>                    | <i>Notable Section:</i><br><i>Disability Overview</i>  | www.hud.gov         |
| <b>National Council of State Housing Agencies (HCSHA)</b>                     | Assist the Housing Finance Agencies' efforts to provide affordable housing to those who need it.<br><br><i>Notable Article:</i><br><i>Special Needs Housing</i>  | www.ncsha.org       |
| <b>Employer Assistance and Resources Network (EARN)</b>                       | EARN provides resources for employment seekers and job training for those looking to gain and build skills necessary to succeed in the workforce.  | www.askearn.org     |
| <b>Financial Assistance and Support Services for People with Disabilities</b> | This government listing provides resources on a wide range of tools for people with disabilities to apply for financial assistance with medical treatment, as well as housing resources and tax help.<br><br><i>Notable Resource:</i><br><i>Financial Assistance and Support Services for People with Disabilities</i> | www.usa.gov         |
| <b>Volunteers of America</b>  | This site contains resources for affordable housing, employment, and behavioral health services.<br><br><i>Notable Resources:</i><br><i>People with Disabilities</i>   | www.voa.org         |
| <b>Ability Jobs</b>   | Works with companies, government and nonprofit agencies to employ people with disabilities.  | www.abilityjobs.com |
| <b>Equal Opportunity Publications. Inc. (EOP)</b>                             | EOP is an online publication that publishes career guidance and recruitment magazines for women, members of minority groups and people with disabilities.  | www.eop.com         |

| Organization                                       | Description  | Website  |
|--|--|--|
| <b>Housing Center for People with Disabilities</b> | <p>This website is managed by the Technical Assistance Collaborative and provides information and news updates regarding affordable housing policies and programs for people with disabilities.</p> <p><i>Notable Sections:</i><br/> <i>State + Local Affordable Housing Planning</i><br/> <i>Rental Assistance</i><br/> <i>Housing-Related Supports</i><br/> <i>Section 8 Made Simple</i></p> | <a href="http://www.tacinc.org">www.tacinc.org</a>                                     |
| <b>Mainstream</b>                                  | <p>A national nonprofit organization dedicated to improving competitive employment opportunities for people with disabilities by providing specialized services and acting as a bridge that links people with disabilities, employers and service providers.</p>   | <a href="http://www.mainstreaminc.net">www.mainstreaminc.net</a>                       |
| <b>Bender Consulting</b>                           | <p>Consulting firm that specializes in recruiting and hiring people with disabilities for full-time, competitive employment opportunities.</p>   | <a href="http://www.benderconsult.com">www.benderconsult.com</a>                       |
| <b>Independent Living</b>                          | <p>The NCIL provides resources regarding independent living and a national directory of Centers for Independent Living.</p>  | <a href="http://www.ncil.org">www.ncil.org</a>   |
| <b>PACER</b>                                       | <p>PACER Center enhances the quality of life and expands opportunities for children, youth, and young adults with all disabilities and their families so each person can reach his or her highest potential.</p> <p><i>Notable Resource:</i><br/> <i>Transitioning to Life After High School</i></p>   | <a href="http://www.pacer.org">www.pacer.org</a>                                       |
| <b>Department of Education</b>                     | <p><i>Notable Resource:</i><br/> <i>Office of Vocational Rehabilitation</i></p>  | <a href="http://www.ed.gov">www.ed.gov</a>   |
| <b>Affordable Colleges</b>                         | <p>Empowers students to choose the right education, at the right place, making higher learning accessible for all. They recognize each student has a unique educational path with different obstacles.</p> <p><i>Notable Resource:</i><br/> <i>College Resources for Students with Disabilities</i></p>  | <a href="http://www.affordablecollegesonline.org">www.affordablecollegesonline.org</a> |

## Section Eleven: Assistive technology (tools and devices)

Assistive tools and technologies are designed to help individuals with life-limiting circumstances, such as an intellectual disability, to better learn, communicate, and function in day-to-day life. These tools can promote inclusion and enable individuals to have greater independence and participation in activities such as school, work and community or civic life. While assistive technology can refer to a wide range of tools or resources, they all offer the potential to enhance the quality of a disabled individual's life.

### What is assistive technology?

Assistive technology means any special device or equipment that helps people with disabilities in daily life. Common examples include electronic communication aids, wheelchairs, hearing aids, and screen readers. Equipment or tools can be high or low tech so understanding the limitations of an individual's disability can help to match them with appropriate accommodations and technologies.

Access and provisions for assistive technology are regulated under the Technology Related-Assistance for Individuals with Disabilities Act, more commonly known as the Tech Act, which expands access to, availability of and funding for assistive technology for all individuals with disabilities. Originally passed in 1988, the Tech Act was permanently authorized under the Improving Access to Assistive Technology for Individuals with Disabilities Act of 2004.

Assistive technology is also included as a part of the Individuals with Disabilities Education Act (IDEA), which provides for specific consideration of assistive technology when developing Individual Education Programs (IEP). Schools are obligated to provide assistive technology at home or in other environments where an IEP team determines that a student needs assistive technology to benefit from his or her educational program. The need for assistive technology must be considered, like other needs, on a case-by-case basis.

Under the Tech Act, an assistive technology device means "any piece of equipment or product or system, whether acquired commercially off the shelf, modified, or customized, that is used to increase, maintain, or improve the functional capabilities of children with disabilities." It further defines an assistive technology service as "any service that directly assists an individual with a disability in selection, acquisition or use of an assistive technology device." (P.L. 108-364)

### What is the purpose/benefit of assistive technology?

Assistive tools and technologies can support individuals within intellectual or physical limitations to participate more actively in many facets of their lives. They can also promote greater independence and reduce the need for caregiver support in certain areas, which enables a better quality of life for individuals and their families.

Assistive technologies can include mechanical, electronic, and microprocessor-based equipment, non-medical and non-electronic aids, specialized instructional materials, services, and strategies that people with disabilities can use either to:

- assist them in learning
- make the environment more accessible
- enable them to compete in the workplace
- enhance their independence or otherwise improve their quality of life

## How do you fund these services?

Under the Tech Act, each U.S. state and territory receives a grant to fund an Assistive Technology Act Project (ATAP). Services may vary by state, but generally will provide the following types of assistance:

- Access to computerized information on the use of assistive technology
- Develop demonstration centers where people with disabilities can try different pieces of equipment
- Consultants to help individuals make informed decisions about equipment
- Referral services
- Training to both individuals with disabilities and others

Depending on the demonstrated need, type of device, tool or technology, and state/local resources, funding and reimbursement for assistive technology can vary widely. In many cases, where an IEP includes specific requirements for these tools, the school system will be responsible for the cost. Outside of IEPs, funding can come from government programs (SSI, Medicaid, etc.), private insurance, employers or local charities or disability support organizations. For additional information, it is recommended to visit the Assistive Technology program in your state to explore what funding or support is offered in your area.

To support the need for assistive technology, parents and professionals should document precisely how the student would benefit educationally. For example, documentation may include a specific example such as: "Mary usually takes 1 hour to do 8 math problems. With a calculator, she can do the same number of problems in 20 minutes." For more information on IEPs, see Section Eight of this guide.

## Seizure-alert dogs

Many of the TSC individuals with epilepsy also have a seizure dog who has been professionally trained to respond to a seizure for that individual. Examples of this training would include barking or alerting family members when a seizure is happening, some are trained to lie next to the individual to prevent injury, and some dogs are trained to use their body to help break the fall of the seizing individual to prevent further injury.

To find out if a seizure-alert dog is the right fit for your situation please refer to the additional resources at the end of this section.

*Please see the TSC Navigator for updated resources on seizure dog organizations.*

## Section Eleven Appendix

| Organization                | Description  | Website  |
|-----------------------------|--|--|
| <b>KidNeeds</b>             | Provides resources for special needs children and their families for different types of disabilities, growth, development, and technology  | <a href="http://www.kidneeds.com">www.kidneeds.com</a>                                     |
| <b>Danny Did Foundation</b> | A resource for families who are impacted by seizures<br><br><i>Notable Section:</i><br><i>Devices + Technology</i>   | <a href="http://www.dannydid.org">www.dannydid.org</a>                                     |
| <b>Epilepsy Foundation</b>  | To lead the fight to overcome the challenges of living with epilepsy and to accelerate therapies to stop seizures, find cures, and save lives.<br><br><i>Notable Section:</i><br><i>Epilepsy Device Wiki</i>   | <a href="http://www.epilepsy.com">www.epilepsy.com</a>                                     |
| <b>Charlie Foundation</b>   | Dedicated to advancing the awareness of ketogenic therapeutics to help with seizures.  | <a href="http://www.charliefoundation.org">www.charliefoundation.org</a>                   |
| <b>Seizure Tracker</b>      | Founded by parents of a child with TSC who were struggling to understand their son's seizure activity, Seizure Tracker™ has become a valuable tool for parents, doctors, and researchers alike.  | <a href="http://www.seizuretracker.com">www.seizuretracker.com</a>                         |
| <b>Chelsea Hutchison</b>    | A non-profit to provide help and support to individuals, particularly children and young adults, who have epilepsy.<br><br><i>Notable Section:</i><br><i>Helpful Links: Monitoring Devices</i><br><i>Helpful Link: Service Dogs</i><br><i>Helpful Links: Other</i>   | <a href="http://www.chelseahutchisonfoundation.org">www.chelseahutchisonfoundation.org</a> |
| <b>4 Paws for Ability</b>   | 4 Paws for Ability enriches the lives of people with disabilities by placing life changing service dogs. We breed, raise, and train our service dogs in preparation to assist their partner with specific tasks and skills that mitigate the impacts of their partner's disabilities. We work hard, love harder, and play the hardest. | <a href="http://www.4pawsforability.org">www.4pawsforability.org</a>                       |



## Questions to ask about canine assistance

Are you an adult or parent of a child with TSC who has epilepsy or other physical disabilities or special needs who is considering getting a seizure response or service dog? Use this list of questions to help you choose a canine assistance program<sup>1</sup> that best fits your needs.

1. "My child is only 4 years old." What is the youngest age you will train a dog for, and will you train a dog for someone with an intellectual disability?
2. What skills can you train the dog to do?
  - a. Example: Seizure training, autism (tethering, tracking), anxiety concerns, behavior disruption, diabetes, hearing impaired, etc.
3. What's involved with the application process and fee?
4. Is there a cost associated to get a dog, if so, how much?
5. What breeds of dogs do you train?
  - a. Are you able to accommodate for persons with allergies to certain breeds of dogs?
6. How long is the waiting list to get a dog?
7. How many dogs do you train a year?
8. Do you offer a guarantee for dog training and what happens if my dog doesn't work out?
9. If my application is accepted, what training is involved for me to ensure a successful pairing with my dog?
  - a. Do I have to travel to your facility?
  - b. How long does my training last?
  - c. Is lodging and food covered?
  - d. What supplies do I need to bring with me?
  - e. Do you offer follow-up training if needed?
10. Do you provide any future veterinary care or referral to a local veterinarian who provides free or reduced cost services for my dog?
11. How old will the dog be when I receive him/her, and how long will the dog remain in active service?
12. Will we be able to adopt the dog after he/she's retired?

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<sup>1</sup> The TSC Alliance does not endorse any canine assistant program. Search "service dogs" or "assistance dogs" on the internet for a list of programs.

## Section Twelve: Resources and opportunities for your child to live his/her best life

When you find out your child is diagnosed with a complex medical condition like TSC with an unpredictable road ahead, it's natural to think about all the things the child may miss out on that his or her peers are doing. We often feel we have been dealt all the wrong cards, which is certainly a valid feeling. It can be hard to find resources and activities suitable for your child. This section will briefly discuss additional therapy options like art and music, sporting activities, social interactions and other extra-curricular activities.

### Social communication

Communication, both verbal and non-verbal, is essential to social interactions. Children often learn communication skills from observing interactions between peers and adults. For some of our kids, communicating with others is hard, stressful and at times can cause additional behaviors, because of this learning social communication usually requires direct modeling and intervention.

### Art therapy

Art therapy is a great way to work with your child's strengths on his/her own terms, working on fine and gross motor skills in a fun and interactive environment. It looks at how art can affect the neurological system and can be seen as an intervention added to a student's IEP. Although special education eligibility alone is not an automatic qualifier for art therapy in the education setting, other factors come into play that may help get art therapy on an individual's IEP. For more information about art therapy, please refer to the resources at the end of this section.

### Music therapy

A great way to improve social and communication skills is through music. Music therapy is another intervention used to help with communication, social, cognition and motor skills. Simple songs can be used to remember daily tasks or transitions in the classroom. Music can help improve communication skills by imitating body movements, language, cooperative play and group participation.

### Dance therapy

Just like with music therapy, dance can also improve motor skills, body coordination and control, body awareness and sensory input/output. Most importantly dance provides other opportunities for individuals to communicate, especially those who are non-verbal or lack communication skills.

### Drama therapy

This form of therapy is great for sensory exploration by using movement, touch, sound and colors to help engage the senses. It also provides different ways for individuals to communicate in a stress-free environment.

### Sports

A part of childhood is playing sports. Regardless of an individual's ability, everyone has the right to enjoy playing sports. Many sports can be adapted or modified to allow those with disabilities to partake in. The thought of 'limitations' should not prevent an individual from enjoying a sport or even trying in the first place.

There has been a growing trend in making sports inclusive to all abilities and offering opportunities to those who would otherwise never have the chance to participate. It is recognized barriers exist to accessing these sports programs for disabled individuals. For more information about different sports programs please refer to the resources at the end of this section.

Just like with any sport, there are limitations to consider. One thing to remember when your child is participating in a physical activity is to make sure an adult who is aware of your child's particular physical limitations is always around. If your child has seizures, make sure this is communicated, including any triggers the staff needs to know about. For those who have children with TSC and PKD, participating in any contact sports is not recommended due to the high risk of rupture of the kidney. As with any child, regardless of having TSC, if they are going to participate in any water activities, make sure swim safety is discussed and training has been done by the staff.

## Section Twelve Appendix

| Organization   | Description   | Website  |
|--|---|--|
| <b>PACER</b>   | <i>Notable Resource:</i><br><i>Inclusive Recreation, Adaptive Sports, and Summer Camps</i>  | <a href="http://www.pacer.org">www.pacer.org</a>   |
| <b>Danny Did Foundation</b>                                    | A resource for families who are impacted by seizures<br><br><i>Notable Section:</i><br><i>Devices + Technology</i>  | <a href="http://www.dannydid.org">www.dannydid.org</a>   |
| <b>American Association of Adapted Sports Programs (AAASP)</b> | AAASP's mission is to oversee the partnership of leaders in education and community to lay the foundation for a national network of interscholastic adapted athletic programs.  | <a href="http://www.adaptedsports.org">www.adaptedsports.org</a>   |
| <b>E-Sports-Exceptional</b>                                    | A fully inclusive program dedicated to empowering kids of all abilities to reach their full athletic and social potential. The "E" in E-Sports stands for "exceptional" because we believe all people, both with typical and special needs, are exceptional.  | <a href="http://www.e-sports.org">www.e-sports.org</a>   |
| <b>Special Olympics</b>  | The Special Olympics is a worldwide organization offering a variety of sports opportunities to individuals with intellectual disabilities. With over 30 Olympic-style sports offered, there are countless opportunities for individuals to participate.   | <a href="http://www.specialolympics.org">www.specialolympics.org</a>   |
| <b>Miracle League</b>  | Even the most disabled individuals deserve a chance to experience play sports. The Miracle League is a great opportunity for those who have mobile disabilities still get a chance to play baseball. Parents or "buddies" accompany the individual on the field, as just an aide and other times assisting them with the bat. | <a href="http://www.miracleleague.com">www.miracleleague.com</a>   |
| <b>Little League Challenger Division</b>                       | Most Little Leagues have a challenger division for individuals with disabilities. The teams are grouped by abilities not age. Teams range from tee-ball, coach pitch, to player pitch.  | <a href="http://www.littleleague.org/play-little-league/challenger">www.littleleague.org/play-little-league/challenger</a> |
| <b>Challenged Athletes Foundation (CAF)</b>                    | It is the mission of the Challenged Athletes Foundation (CAF) to provide opportunities and support to people with physical challenges, so they can pursue active lifestyles through physical fitness and competitive athletics.   | <a href="http://www.challengedathletes.org">www.challengedathletes.org</a>   |

| Organization   | Description  | Website  |
|--|--|--|
| <b>KEEN (Kids Enjoy Exercise Now)</b>  | KEEN is a volunteer-led, nonprofit organization headquartered in the Washington, DC area. KEEN's mission is to provide free programs for kids with disabilities, regardless of the nature or severity of that disability.  | <a href="http://www.keenusa.org">www.keenusa.org</a>                                 |
| <b>Sports for Kids Foundation</b>  | The Sports for Kids Foundation is dedicated to improving the quality of life of special needs children and their families throughout the United States.  | <a href="http://www.sportsforkidsfoundation.com">www.sportsforkidsfoundation.com</a> |
| <b>AAPM&amp;R Directory of Sports Organizations for Athletes with Disabilities</b> | This directory contains resources for athletes with disabilities who wish to engage in sports competitions and activities and for physiatrists and the general public who would like to volunteer with these organizations.  | <a href="http://www.aapmr.org">www.aapmr.org</a>                                     |
| <b>Bounce out the Stigma</b>   | The primary charter of Bounce Out the Stigma is to empower special needs children and young adults with motor skill sets, self-confidence, peer support, and a unique message to meet the challenges they face. We have carefully constructed our entire platform of programs and events to meet the changing landscape of special needs kids.   | <a href="http://www.bounceoutthestigma.org">www.bounceoutthestigma.org</a>           |
| <b>Pop Warner Challenger Division</b>  | The philosophy of the Pop Warner Challenger Division is to provide the framework for local Pop Warner programs to offer a structured football program for participants with special needs. The Challenger program is non-competitive, and no score is kept. The games are modeled after a typical Pop Warner game with warm-ups, coin toss, and national anthem, etc. Participation in the Challenger Division is permitted at the request of the individual's parent or guardian. | <a href="http://www.popwarner.com">www.popwarner.com</a>                             |
| <b>American Art Therapy Association</b>  | <p>The American Art Therapy Association is a not-for-profit, non-partisan, professional, and educational organization dedicated to the growth and development of the art therapy profession.</p> <p><i>Notable Resource:<br/>About Art Therapy</i></p>   | <a href="http://www.arttherapy.org/">www.arttherapy.org/</a>                         |
| <b>American Music Therapy Association</b>  | <i>Notable Resource:<br/>Music Therapy in Special Education</i>  | <a href="http://www.musictherapy.org">www.musictherapy.org</a>                       |

## Section Thirteen: Life and financial planning

### Financial and estate planning

What is estate and financial planning? This topic can seem overwhelming with the breadth of information available. Estate and financial planning typically go hand-in-hand, but they are distinct from each other. This section will help you get familiar with the process and prepare you to dive in deeper.

- **Financial planning** focuses on addressing long-term financial goals. Examples of financial planning include ABLE accounts and Special Needs Trust. A skilled financial planner can help customize your unique goals while mapping out a financial road map to achieve your goals.
- **Estate planning**, when conducted with the assistance of an estate planning attorney, helps protect your assets. An ideal estate plan should include a trust and living will.

### Achieving a Better Life Act (ABLE)

The ABLE account allows families to create tax-free saving accounts that would cover qualified expenses associated with the individual's disability without losing eligibility for public benefits.

### Special Needs Trust (SNT)

A SNT is a trust that will preserve the individual's eligibility for government-based programs such as Medicaid and Social Security Income (SSI). This happens because the individual, or beneficiary, does not own the assets in the trust, he/she remains eligible for the benefits. The trustee of the account is able to supplement the beneficiary's government-based benefits but not replace them. An example of these supplemental needs includes sitters, companions, dental and medical expenses that are not covered by either Medicaid or Medicare.

### Life and future planning

This section will help you get familiar with the process and prepare you to dive in deeper.

Estate and financial planning typically go hand-in-hand, but they are distinct from each other. Financial planning focuses on addressing long-term financial goals. A skilled financial planner can help customize your unique goals while mapping out a financial road map to achieve your goals. Estate planning, when conducted with the assistance of an estate planning attorney, helps protect your assets. An ideal estate plan should include a trust and living will. Estate planning may also include guardianship and conservatorship.

It is never too early to start planning for your or your loved one's future. Like TSC, all estate and financial journeys are unique. Some trusts could potentially affect you and your loved one's eligibility for benefits (e.g., Medicaid, Social Security Income, etc.), while others can be accessed for service reimbursement, leaving you or your loved one without a secure financial future. Having a firm understanding will help empower you to initiate the process and examine essential questions to help guide and reach your future goals, giving you peace of mind when you are no longer available to care for your loved one. MassMutual SpecialCare is a dedicated program that will create a holistic plan that goes beyond typical financial matters.

### Guardianships

A guardianship – or conservatorship depending on the state – is a legal action that grants an adult legal power to make decisions for another person. Guardianship is a legal means for protecting adults who cannot take care of themselves, make decisions that are in their own best interest or handle their assets.

Generally, the natural guardianship of a minor child terminates at the age of 18, and the parent is no longer the child's legal guardian regardless of a disability. Some adults with disabilities may not be able to give reasoned and well-informed consent when making a decision. Depending on the severity of the adult's disability, setting up a guardianship may be an option.

However, keep in mind guardianships can be relatively inflexible as compared to less intrusive options such as trusts (described later in this section). When considering the different types of available financial planning tools, families will need to consider some of the following questions:

- Is the individual able to work? If so, to what extent?
- Is the individual able to manage small amounts of money on a monthly basis?
- Does the individual now or in the future need residential care?
- Can the individual live with a friend, relative or group home in the future?
- What are the estimated costs of these arrangements?
- What are the person's recreation, leisure time and social needs?
- Does the person's disability involve the possibility of deteriorating health and more involved health care needs and costs?
- What will the transportation costs for this individual be now and in the future?

The following is a brief description of the types of guardianships generally available:

- A Guardian of the Person is responsible for monitoring the care of the ward.
- A Guardian of the Estate or Conservatorship should be considered for persons with disabilities who are unable to manage their finances and who have income from sources other than benefit checks or have other assets and/or property.
- A Limited Guardianship may limit the guardian's decision making to certain areas, such as decisions about medical treatment, to allow the ward to continue making his/her own decisions in all other areas.
- A Temporary Guardian or Conservator may be appointed in an emergency when certain decisions must be made immediately.

A **letter of intent** is an important accompanying document for guardianships. A letter of intent describes the person's disability history, his/her current state and what future needs may be. This document is important as it provides guidance to the trustee regarding the family's wishes for the family member with TSC in the future. To the maximum extent possible, involve the adult in the writing of this letter, so the letter truly represents his or her interests. Completing the letter of intent should be done as soon as possible and updated regularly to reflect any changes in a person's health status or situation. This ensures the letter is ready at any moment should a parent become ill, become disabled or die. Although it is not legally binding, it provides direction for the person(s) who will care for the adult with special needs in the future.

## Alternatives to guardianships

Parents, family members and/or other potential caregivers must carefully consider the disabled adult's individual circumstances, including strengths/weaknesses, needs and interests, before deciding to seek guardianship. If the adult with a disability is capable of making some but not all decisions, some less-intrusive alternatives to guardianship listed below may be considered:

- A **Representative Payee** (often a family member, friend, or nonprofit agency) can be named to manage the funds of a person with a disability who receives government benefits checks, such as SSI and SSDI.
- A **Durable Power of Attorney for Property** is useful for individuals with mild or moderate disabilities who are capable of choosing another person to handle their money.
- A **Durable Power of Attorney for Health Care** or **Medical Power of Attorney**, also known as a **Health Care Proxy**, should be considered for individuals who are disabled and who can make some, but not all health care decisions. This is a legal document that enables a competent individual (the "principal") to designate a health care agent to make health care decisions should the individual become incompetent to make them.
- An **Appointment of Advocate and Authorization** is a customized power of attorney that allows an individual with a disability to designate an agent to advocate on his/her behalf with administrative agencies such as the Department of Developmental Services, the Department of Human Services, Medicaid, local education authorities and any other state or federal agency from which an individual is receiving services.

As individuals and their families consider the different legal planning tools that will work best for their situation, keep in mind the Social Security Administration (SSA) will only speak with a representative payee with regard to a person's benefit checks and will not speak with an agent who has been designated as a durable power of attorney for health care or advocate for the individual with a disability.

## Section Thirteen Appendix

| Organization                          | Description   | Website  |
|---------------------------------------|---|--|
| <b>Special Needs Alliance</b>         | <p>National alliance of attorneys for special needs planning.</p> <p><i>Notable Resource:</i><br/> <i>Life Care Planning</i><br/> <i>Special Needs Trust</i><br/> <i>ABLE Accounts</i></p>  | <a href="http://www.specialneedsalliance.org/">www.specialneedsalliance.org/</a> |
| <b>Social Security Administration</b> | <p><i>Notable Resource:</i><br/> <i>Spotlight on Achieving a Better Life Experience (ABLE)</i></p>  | <a href="http://www.ssa.gov">www.ssa.gov</a>                                     |
| <b>Mass Mutual</b>                    | <p>A Mutual Life Insurance and Financial Service Company.</p> <p><i>Notable Resource:</i><br/> <i>SpecialCare</i></p>   | <a href="http://www.massmutual.com">www.massmutual.com</a>                       |
| <b>ABLE National Resource Center</b>  | <p>Connecting people with disabilities, their families and those who support them to information about ABLE</p>   | <a href="http://www.ablenrc.org">www.ablenrc.org</a>                             |
| <b>National Guardianship</b>          | <p>Its mission is to establish and promote a nationally recognized standard of excellence in guardianship. Membership provides access to the NGA newsletter, use of the ethics hotline and extensive links for guardianship management.</p> | <a href="http://www.guardianship.org">www.guardianship.org</a>                   |
| <b>Future Planning</b>                | <p>This resource list from The Arc includes government agencies, publications regarding estate planning, guardianship financial and future planning resources by state.</p> <p><i>Notable Resource:</i><br/> <i>Future Planning</i></p>     | <a href="http://www.thearc.org">www.thearc.org</a>                               |
| <b>National Law Review</b>            | <p>The National Law Review is a free to use, no-log in database of legal and business articles.</p> <p><i>Notable Article:</i><br/> <i>Understanding Special Needs Trusts</i></p>   | <a href="http://www.natlawreview.com">www.natlawreview.com</a>                   |

## Section Fourteen: Self-care and family dynamics

### Self-care and mental health

Being diagnosed with TSC can cause significant challenges within the life of the individual regardless of when first diagnosed. It is essential to understand acknowledging that life is different can be difficult for some while for others it might seem relieving to finally have a “name” to the issues they’ve experienced for several years.

It is completely valid and okay to have mixed emotions and process the reality that everyone’s “normal” is no longer the same. Another important factor to consider is that despite the wide range of neurocognitive abilities for those with TSC, discrimination can appear different as the individual gets older. The emotional challenges can be just as debilitating as physical challenges and significantly trigger ineffective coping mechanisms, stress, and anxiety.

### LGBTQIA+ community and TSC

The TSC Alliance supports people with TSC who are LGBTQIA+; therefore, the unique and unfortunate challenges experienced in this community are just as important to address as all other barriers and challenges along this journey. Many will face the triple discrimination of living with a genetic disorder, coping with a TSC-associated neuropsychiatric manifestation and grappling with gender identity all greatly impact the level of stress, social stigma and discrimination compared to cisgender peers with TSC. It is essential to reinforce that identifying as LGBTQIA+ is not a mental health illness.

Sadly, many LGBTQIA+ individuals experience significant disproportionate mental health struggles including suicide. We acknowledge that exclusion, access to care and lack of safety create additional barriers to your overall health and wellbeing. In addition to the above recommendations for self-care surrounding mental health, make sure you have the right healthcare team to not only address your TSC needs but your personal and physical wellbeing too.

The GLMA Provider Directory is the recommended resource to locate a LGBTQI+ friendly provider to help manage your overall healthcare needs. Your safety and the ability to feel comfortable sharing your identity with family and friends can be difficult, which is why we recommend The Epilepsy Foundation’s Epilepsy & Seizure Helpline in English (1-800-332-1000) and Spanish (1-866-748-8008), which is a free, confidential helpline available 24 hours, 7 days a week. Additional mental health resources for LGBTQIA+ individuals are available in the appendix section. We also encourage you to contact one of our TSC Navigators if you have any questions or concerns about navigating your journey.

### Caregiver mental health

Being a caregiver can be equally rewarding and overwhelming at the same time – it is completely normal to feel this way. Many caregivers start this journey as parents, yet others might be family relatives (grandparents, siblings, etc.) or close family friends who are all essential parts of the “village” that cares for someone with a disability. Western society often considers the village as solely for the loved one who needs assistance; however, the reality is that the village is the support system for the primary caregiver in addition to the person with a disability.

Regardless of where you are on this journey it is important to take a moment and realize the first step in acknowledging caregiver mental health is accepting that grief started at diagnosis. It is completely valid and okay to have mixed emotions and process the reality that everyone is “normal” is no longer the same. Many caregivers throw themselves into understanding the disease, researching treatment options, experts, and resources for their loved one, yet most do not take the same advice on finding those same resources, tricks, and tools to help them become the best caregiver they can become. This new normal might not be what you expected but you can still have a sense of control even under the most difficult circumstances.

Caring for a loved one with a rare disease such as TSC means having to work especially hard at finding balance. It is all too common for caregivers in our community to find themselves without a sense of balance in their lives. Too often, caregivers will find themselves exhausting their mind and body in an effort to provide the absolute best for their loved one while neglecting their own basic needs.

In the midst of being overwhelmed, it can also be challenging to even know where to start, how to recognize if you are close to “burning out,” and what action steps you can take to create healthy boundaries so you can live your fullest life. Below are some signs and symptoms of caregiver burnout. If you or another caregiver is experiencing these symptoms, we recommend reaching out to your local provider or therapist to work through your unique situation and provide you with plan that meets your needs.

The TSC journey can often bring ups and downs given the uncertainties that exist during non-conflict, or non-crisis times, understanding your Caregiver Zone can help you gauge the amount of stress you are currently under. Stress levels constantly ebb and flow – you may have seasons where you fluctuate between the different Caregiver Zones. This is normal and to be expected. Once you have had time to implement this you start to uncover trends in your life and be able to start proactively empowering strategies that work for you and your family dynamic. It is not about surviving this life; it is about uniquely thriving the best you can with the resources you have.

Another resource is the How to Help Me Guide where you will find a list of examples that you can customize to fit your own needs. As caregivers going through challenging times you often hear, “how can I help you.” Yet, you might not even know that because you are already in a crisis and decision fatigue is already setting in. Having a go to list that you can show to others not only helps you but helps those in your inner circle to know how to truly help you. Locate the Finding your Caregiver Zone and How to Help Me guide and worksheet in the appendix of this section for additional support. Please visit the TSC Navigator periodically for added resources and updated content.

### Signs and symptoms of caregiver burnout

If you find yourself identifying with a lot of these feelings, please bring this sheet and reach out to your local PCP or local mental health provider.

#### Physical

- No energy, “run down” feeling
- Changes in your body weight
- Chronic or acute pain and fatigue
- Sleep cycle disturbances
- Nausea (with or without vomiting)
- Changes in bowel movements (diarrhea or constipation)
- Frequent cold-like illness or congestion

#### Emotional

- Mood Swings and/or stronger-than-usual emotions (anger, crying, etc.)
- Emotional numbness
- Lack of focus/concentration
- Constant state of feeling overwhelmed or worried
- Frequent emotions of guilt and/or resentment

#### Behavioral

- Neglecting your own basic health needs (appointments, screenings, hygiene)
- Not enjoying the things that you used to
- Avoiding your friends and family
- Detachment from loved ones
- Increased use of alcohol, drugs, and/or prescription medications

***If you feel an immediate threat to yourself or others, please call 911 for immediate assistance. If you are having suicidal thoughts, please call the National Suicide Prevention Lifeline at 1-800-273-8255.***

### Family discussions

Discussing a new diagnosis with your family is also a process that can be challenging during this phase. Each situation is unique and disclosing personal information with friends and family should occur only when you are ready to have that discussion. Because this part can be difficult, the TSC Alliance crafted a letter you can share with friends and family to help explain the diagnosis when you might not have yet found the words to say. You can find this letter in the Tips from Parents section of the TSC Navigator as well as the appendix of this section.

## How to talk to your child about TSC

The right time to tell your child he or she has tuberous sclerosis complex will depend on family dynamics, your child's neurocognitive abilities and maturity level. During this age group it is important to provide stability and balance where possible. As research and technology has improved, more individuals are being diagnosed during prenatal screenings and evaluations. Working with your healthcare team including child life specialists and therapist, learning about TSC will happen gradually over the years. Several resources — such as books *Princess Katie the Brave: A Story About Living with TSC*, *Teddie Rocket: The Astronaut with TSC*, and *My Life with TSC* — are available to help normalize the complexity of their TSC Journey. You can find these books on Amazon.

As your child grows, this conversation will adapt to the psycho-social aspects of having chronic disease. However, during this age group, providing simple yet truthful statements to help foster and healthy curiosity with balancing to eliminate fears. There is no right or wrong answer on when you should tell your child on their diagnosis. If you feel like you are struggling with this, please contact your local medical team for additional resources.

## How to talk to your unaffected child(ren) about TSC

Siblings of children with disabilities face mixed challenges that all individuals within the family dynamic experience across this journey. Older siblings can often feel responsible for their younger sibling's disability/diagnosis while others feel protective, embarrassed, confused and even resentful. Younger siblings do not know life any different. They can typically be more curious and accepting of the situation simply due to the birth order, but as they grow, they often feel very protective and the "guardian" of their older sibling. It's essential to acknowledge that what they display outwardly does not mean internally they have different emotions. Depending on the age gap, it's important as the guardian to understand that all emotions are normal. Adding additional siblings is a significant life change, and when one child requires more time from you, it's normal for your unaffected child(ren) to have a hard time adjusting.

The most vital thing you can do is give your unaffected child(ren) consistent individual time with them. Let them express their feelings. It's okay if their emotions do not match with the reality of the situation, but with consistent individual time together, improved coping and communication skills will develop. It is also important to set a healthy example and verbalize it's okay to ask for help. It's acceptable for things to feel like they are not okay and verbalizing it is important.

Another key factor is explaining the disability to your unaffected child(ren) at their age-appropriate level of understanding, honestly as possible. It is essential to meet your unaffected child(ren) where they are at each milestone, emotion and significant event that occurs throughout this journey, which like your affected child, will change over the years. Providing peer support and professional assistance can also help your unaffected child(ren) develop the skills as they too are on this journey. Another helpful option is to help your unaffected articulate what to say when things might come up in public or at school. Helping them proactively have problem solving skills can help eliminate barriers and issues on their TSC journey.

Regardless of your journey, ensuring your unaffected child(ren) feels needed and equally important is essential to establish a healthy understanding and strong family dynamic. Consistently show up and be present. Set boundaries together that during their individual time it's their time. And if/when you cannot show up, remember you are teaching them by example how to communicate and adjust to life challenges. It's okay to teach them how to work through challenges. You're going to mess up, and that's okay. It's what you do next that will leave an everlasting impression. Be accountable. Be honest. Be raw. Be you.

## Respite care

For families of a child with a disability, respite care is a support service that may be required from time to time. There are several types of respite care, but the two main models of respite care are in-home and out-of-home options. Some community-based programs offer respite care that may be as simple as providing a substitute caregiver for a few hours each week. Others provide more major interventions, like temporarily placing the individual in a special respite home. For example, some Epilepsy Foundation affiliates provide respite care for individuals who experience frequent and debilitating seizures. Care is typically provided for an agreed upon time.

To find out if there is an affiliate in your community that provides this service, visit the Epilepsy Foundation at [www.epilepsy.com](http://www.epilepsy.com). In addition, the National Respite Locator Service helps individuals, parents and caregivers find respite services in their state and local area.

To learn more about types of respite care please visit TSC Navigator: ABCs of Respite: A Consumer Guide for Family Caregivers.

*\*Note that respite care and services vary by state and region.*

## Section Fourteen Appendix

| Organization  | Description  | Website  |
|---|--|--|
| <b>Child Neurology Foundation</b>   | <p>To serve as a collaborative center of education, resources, and support for children and their families living with neurologic conditions and facilitate connection with medical professionals who care for them.</p> <p><i>Notable Articles:<br/>Tools &amp; Resources<br/>Preparing for Your Doctor's Visit<br/>Support for Patients &amp; Caregivers</i></p> | <a href="http://www.childneurologyfoundation.org">www.childneurologyfoundation.org</a> |
| <b>Charlie Foundation</b>   | <p>Was founded in 1994 to provide information about diet therapies for people with epilepsy, other neurological disorders and select cancers.</p> <p><i>Notable Article:<br/>Ketogenic Therapies &amp; Resources</i></p>   | <a href="http://www.charliefoundation.org">www.charliefoundation.org</a>               |
| <b>Variety</b>  | <p>Variety-The Children's Charity serves children who may fall through the cracks of government funding or other aid. Depending on government policy and the availability of other assistance, items and services granted by Variety do vary from state to state.</p>  | <a href="http://usvariety.org/about-us/">usvariety.org/about-us/</a>                   |
| <b>Special Needs Project</b>  | <p>Provides books about disabilities, that serve individuals, families, and professionals with a large collection of disability-related materials, including titles about Autism, ADHD, Independent Living and Full Inclusion.</p>   | <a href="http://www.specialneeds.com">www.specialneeds.com</a>                         |
| <b>The M.O.R.G.A.N. Project</b>   | <p>The M.O.R.G.A.N. Project focuses on awareness and support of families and parents caring for special-needs children. They focus on providing families with specialized programs for children with special health care needs, disabilities, sensory issues, and concerns, to improve family quality of life.</p>   | <a href="http://www.themorganproject.org">www.themorganproject.org</a>                 |
| <b>Courageous Parents Network</b><br><br><i>Languages available:<br/>English, Spanish</i> | <p>Empowers, supports and equips families and providers caring for children with serious illnesses.</p>  | <a href="http://www.courageousparentsnetwork.org">www.courageousparentsnetwork.org</a> |

| Organization                                      | Description  | Website  |
|---|--|--|
| <b>Parents Helping Parents (PHP)</b>              | Supports, educates, and inspires families and the community to build bright futures for youth and adults with special needs.   | <a href="http://www.php.com">www.php.com</a>                           |
| <b>Association for Successful Parenting</b>       | A nonprofit dedicated to enhancing the lives of parents living with learning difficulties and their families through education, advocacy and support. It brings together researchers, practitioners and self-advocates to build the capacity of communities and families to support and improve outcomes for parents with intellectual disabilities. | <a href="http://www.achancetoparent.net/">www.achancetoparent.net/</a> |
| <b>The National Alliance for Caregiving (NAC)</b> | Dedicated to improving quality of life for friends and family caregivers and those in their care, by advancing research, advocacy, and innovation. Resources specific to rare disease caregivers can be found under Special Populations and Guidebook Sections.  | <a href="http://www.caregiving.org">www.caregiving.org</a>             |
| <b>Parent to Parent USA</b>                       | Provides emotional and informational support to families of children who have special needs.   | <a href="http://www.p2pusa.org">www.p2pusa.org</a>                     |
| <b>Our Place</b>                                  | A community where you can learn from others, connect with others in your same situation and find services.   | <a href="http://www.joinourplace.com">www.joinourplace.com</a>         |
| <b>Child Mind Institute</b>                       | <p>Dedicated to transforming the lives of children and families struggling with mental health and learning disorders by giving them the help they need.</p> <p><i>Notable Resources:</i><br/> <i>Transgender Kids and Gender Dysphoria</i><br/> <i>Mental Health Challenger of LGBTQ+ Kids</i><br/> <i>Gender and Sexuality</i></p>                  | <a href="http://www.childmind.org">www.childmind.org</a>               |



## Finding Your Caregiver Zone

This chart can help you identify where you need help. You'll find yourself moving between zones, which is completely normal. Track this monthly and keep track and share this with close friends and family so they too can recognize signs of burnout.

|                  | Thriving   | Surviving  | Struggling   | Crisis  |
|------------------|--|--|--|---|
| Emotion          | Emotions are steady with only minor swings.                          | Increased emotional fluctuations – nervous, sad, anxious, etc.   | Constant thoughts of intense emotions – fear, panic, sadness, anger, hopelessness, etc.          | Unable to regulate any emotions. Numbness, complete loss of control, easily aggressive or emotional.  |
| Focus            | Able to focus on and execute plans.                                  | Inconsistent focus but still able to execute plans.  | Decision fatigue, unable to focus or make decisions.   | Extreme inability to focus on any decisions for yourself or others.   |
| Social           | Able to communicate effectively and adjust to changes within reason. | Difficult to communicate and adjust plans, easily overwhelmed, irritated or emotional.                         | Increased difficulty with communication and adaptation. Avoidance of friends, family, work, etc. | Unable to communicate or adapt. Frequent isolation from others – emotionally and/or physically.   |
| Appetite         | Normal appetite.   | Inconsistent appetite.   | Emotional eating or restriction.   | Out of control eating/restriction.  |
| Sleep            | Normal sleep habits.   | Trouble falling asleep or staying asleep.  | Frequent restlessness and disruption of sleep (more often than not)                              | Complete inability to sleep with restlessness and disrupted sleep nightly.  |
| Daily Activities | Feeling consistent with daily performances.                          | Inconsistent with daily performances but still able to achieve most on your "to-do" list. No time for hobbies. | Increased inconsistency, more frequently unable to achieve daily tasks, no time for hobbies.     | Complete inability to get any daily tasks done. Withdrawn from friends/family and tasks/hobbies.  |
| Self-Care        | Providing daily self-care.   | Inconsistent but still providing self-care a few days a week.  | Increased inability for self-care, except for very randomly throughout a month.                  | Inability to perform any self-care or meet basic needs to oneself or others. Self-medicating with drugs, alcohol, prescription medications, food, or other stimuli. |

Recommendations and table adapted from: (1), (2), (3).

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## How to Help Me: Worksheet

After determining which Caregiver Zone, you are in, use the suggested prompts to help you get started. Personalize to how this will be specific to your needs.

|                  | Surviving   | Struggling  | Crisis  |
|------------------|---|---|---|
| Emotion          | Reach out to someone close to schedule regular phone call or lunch. (A person you feel safe with in case you need to discuss life situations) | Reach out to someone close to discuss. Counselors, clergy, primary care providers are also helpful resources)   | Reach out for immediate assistance. Develop a crisis plan with a friend for this type of scenario.<br>Counselor, clergy, Suicide Hotline, or seeking emergency care.                              |
| Focus            | Set a daily goal to stay focused. (There are APPs for that!)  | Can you help me make a task list and prioritize my tasks?   | Can you help me by make a plan for daily check ins with myself or someone else?   |
| Social           | Participating in organization groups or focus groups. Plan one event per week that is for YOU.  | Can you watch my special needs child so I can take my other kid out to do something fun?  | Discuss strategies and resources with a close friend or group to help examine priorities. Do not overcommit. Prioritize   |
| Appetite         | Can you come grocery shopping with me this week?  | Can you help me plan my meals/grocery shop for me?  | Can you help me make sure I have dinners made for the next _ nights?  |
| Sleep            | Can you watch X on this night so I can stay at a local hotel for respite care?  | Take a shower or try to meditate.   | Have friends clear your nighttime responsibilities for you a few nights so you can sleep.   |
| Daily Activities | Create a list of grounding activities like walking, painting, rocking, music etc.   | Ask a friend to help with a chore or task to help focus on priorities.<br>Ask for help with laundry.<br>Can you pick up these prescriptions at this pharmacy? | Can you please help me run x, y, and z errands? [The more specific you are, the better – would a clean kitchen help clear your mind? Or picking up groceries? Taking another kid to the dentist?] |
| Self-Care        | Can we do an at-home spa day?   | Can you absorb some of my responsibilities for 20 minutes 3 days a week so I can take a luxurious shower, make a workout class, meditate.                     | Can you come over at x time and do y so I can take a shower, meditate, take a nice walk with my dog, etc.?  |



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Dear Family Member:

I'm writing because a member of your family has been diagnosed with tuberous sclerosis complex, or TSC.

TSC affects everyone differently, which makes it challenging to understand the disease and to know what to be concerned about and what not to worry about. TSC affects some people severely, while others are so mildly affected, they often remain undiagnosed. Some people with TSC may experience developmental delays and intellectual disability. However, many people with TSC live independent, healthy lives enjoying challenging professions such as doctors, lawyers, educators and researchers.

At least two children born each day in the United States will have TSC. Current estimates of newborn babies affected with TSC are 1 in 6,000. Nearly 1 million people worldwide are known to have TSC, with approximately 50,000 in the United States. There are many undiagnosed cases of TSC due to the obscurity of the disease and the mild symptoms that occur in some people. TSC is as common as amyotrophic lateral sclerosis (Lou Gehrig's Disease) or Duchene's muscular dystrophy but is virtually unknown by the general population.

TSC is caused by mutations in one of two genes: TSC1 or TSC2. Genetic testing is available that can identify the mutation that causes the disease in 85 - 90% of people with TSC. This can be useful for confirming diagnosis and for family planning purposes. If a causative mutation is found in you or a family member, you should consider talking with your doctor and/or meeting with a genetic counselor to discuss these issues further.

TSC is transmitted either through genetic inheritance or as a spontaneous genetic mutation. Two-thirds of TSC cases are the result of spontaneous mutation, meaning neither parent carries a mutation. However, one-third of TSC cases are inherited from a parent. Children have a 50% chance of inheriting TSC if one of their parents has this condition. A parent with a mild case of TSC can produce a child who is more severely affected. In fact, some people have such mild cases they may only find out they also have TSC after their more severely affected child is diagnosed.

TSC can cause the growth of non-malignant tumors in various organs and leads to an increased risk of epilepsy, autism, and cognitive or developmental delay. However, most people with TSC have many, but not all, of the possible manifestations. Family members of infants diagnosed with TSC should particularly watch for signs of infantile spasms, a

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particularly dangerous form of epilepsy that leads to cognitive impairment and delays intellectual and motor development.

The tumors resulting from TSC are non-malignant but may still cause problems. Tumors that grow in the brain are generally diagnosed in infancy or childhood and can block the flow of fluid in the brain. Heart tumors, called cardiac rhabdomyomas, may be found by ultrasound during pregnancy or soon after birth, and they usually shrink during infancy. Tumors in the kidney (called renal angiomyolipomas) are generally diagnosed in adulthood and can lead to bleeding or loss of kidney function.

In addition, women with TSC are at risk of developing a lung condition known as lymphangioleiomyomatosis (LAM), although LAM can rarely occur in men, also. Tumors on the skin, known as angiofibromas, often appear during childhood and adolescence and can be disfiguring and bleed easily if scratched.

Fortunately, treatment options for TSC have expanded greatly in the last decade. Afinitor (everolimus)<sup>®</sup> is a drug that shrinks and stops the growth of tumors and is now FDA-approved for the treatment of subependymal giant cell astrocytomas (SEGAs) in the brain, angiomyolipomas in the kidney and as an added treatment to other antiseizures medications for some individuals with TSC. However, in some cases surgery may be required to remove particularly threatening tumors. Sabril (vigabatrin)<sup>®</sup> and Acthar<sup>®</sup> Gel are approved for the treatment of infantile spasms. Epidiolex (cannabidiol) is approved of seizures associated with TSC. Additionally, we are learning more each day about better ways to treat epilepsy, angiomyolipomas, angiofibromas, and other aspects of TSC.

If you would like to connect with a doctor or TSC Clinic in your area, do not hesitate to contact the TSC Alliance. The TSC Alliance is the only national voluntary health organization dedicated to finding a cure for TSC while improving the lives of those affected. Much more information can be found on our website at [tscalliance.org](http://tscalliance.org).

I hope this information is helpful to you. Should you have any questions or would like to get more information, please contact the TSC Alliance at (800) 225-6872.

Sincerely,

Ashley Pounders MSN, FNP – C  
Director, Medical Affairs  
[apounders@tscalliance.org](mailto:apounders@tscalliance.org)

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## Section Fifteen: Difficult conversations

### Continuum of grief

Grief can accompany many life events big and small; the loss of something that was very important to you is an impactful experience. While it might seem out of place to discuss grief in this guide, the reality is everyone on this journey has experienced grief in their own unique ways. While it is of course natural for parents to focus primarily on the wellbeing of their adult child, it is important to acknowledge the life you once had has forever changed, even if you have been on this TSC journey for decades. Your routines, connections and friendships may change – of course this is a common part of any rare disease journey. Your friends and family might not be aware of TSC, might not understand how to talk to you about this new diagnosis and may want to avoid being intrusive or burdensome.

Reflecting on these pivotal, life-changing moments, it is important to increase awareness to those in your circle who care yet are not quite sure how to even start the conversation. One thing we know well at the TSC Alliance is TSC individuals and families thrive when given unconditional support. So, let us start with where you are.

The utmost important factor to remember is you will cycle through a continuum of grief. Many of you on this journey experienced this years ago from new diagnosis, to failed milestones, to grieving the loss of a significant life event such as kindergarten, field trips, birthday parties, driving, graduation or even future children. Those of you at this point in the journey see these reflections reappear during “the empty nest years” or the experience of having grandchildren. Others on this adult journey might not identify with the grief continuum at an early age; however, grief is still a continuum within this diagnosis journey. We encourage you to discuss with a licensed therapist if you are having a hard time processing this new journey you did not ask to be on.

Some milestones are easier to digest, and others are much harder and downright painful to process. For those you consider to be in your inner circle – your allies – it is important they be mindful of these events. Which event stings the most might not always be clear to you or your close friends, nor will it always be predictable. Allow yourself time to process and express your emotions without judgment or a timeline. Grief never goes away, and your inner circle should avoid making statements that put a timeline on when you should be “over it” or belittling statements about the missed milestone. It is okay to sit with raw sadness. The inability to properly process grief can lead to caregiver burnout and significant mental health issues. Do everything you can to proactively have friendships in place to help be a guardrail for these signs and symptoms.

It is also okay to have your inner circle of friends educated regarding how you feel and how they can help you. For example, you might want to share with them some notes:

*Silence does not have to be awkward. Be mindful and try not to talk during these moments to suggest “fixes” or make light of the situation. Your non-judgmental presence might be all I need. Don’t listen to respond, listen to understand and learn. Examine your own feelings of the situation and really process your own emotions. If you are the friend who has chosen to walk this journey with me, you too must adapt and grow along the way.*

*I do still want to be present in your life and want to hear about your family and even if nine times out of ten I say no to an activity, please keep inviting me. Even if you want to share a milestone that might be triggering for me, please share it (tactfully) so I can celebrate with you. I might not share the same energy and enthusiasm, but I cherish being a part of your life as well.*

And when it comes to helping your friends build relationships with you or your loved one, help them be authentic. Teach them the best ways to engage you. Help your friends learn how they might be able to support you – maybe they can learn about seizure safety or how to administer emergency medication. They may not become experts in TSC, but they certainly can become experts in being your friend.

It’s not an easy journey, but it is your journey, and one you did not choose. Repeat that last line: you did not choose this journey.

## Grief and bereavement

All caregivers should prepare for the possibility of grief or bereavement during the care of a loved one. Caregivers naturally become deeply emotionally invested in the individuals for whom they provide care due to the nature of the position. Serving as a caregiver causes a range of emotions including feelings of happiness and grief throughout the lifespan.

Many caregivers experienced grief starting at the time of TSC diagnosis; grief is not a linear process but a circular one. Often the caregiver travels through a grieving process that leads to a baseline state of worry, stress and frustration. However, even during this process, there will be times of excitement, joy, and immense gratitude that mixes with the grief – creating a blended “chaotic comfort zone.” In the time of the loss of a loved one, it is often difficult to accept moving from the chaotic comfort zone to bereavement and once again having to redefine expectations for short- (daily) and long-term life. This adjustment often requires reevaluation of personal identity and worth.

As you continue to heal from your loss, it is typical to shift from focusing on how your loved one died to focusing on how much he/she lived. This leads to a process in bereavement where you recognize even in personal loss, your bond with your loved one is to be remembered and cherished. Often, the caregiver will identify a path forward that honors the loss of a loved one.

Regardless of the nature of the death, if it was abrupt or expected, you are not alone. Grief is possible at any time and without warning. There is no guide to the grieving process because it is unique to a person. It is important not to drown in others’ expectations of how you should behave. In fact, it is crucial to not let others define how you process grief; you are the architect of your own journey.

If you feel like you don’t know where to start or would like to connect others who have been down this road before, seek support from other special needs caregivers and families. Hearing about how they coped with this difficult transition and what worked for them in the healing process may help guide your own journey.

If you or someone you know is going through grief or bereavement, please contact one of our TSC Support Navigators for additional assistance.

## SUDEP

SUDEP stands for sudden unexpected death in epilepsy. It is a rare but fatal complication of epilepsy with no other explained cause of death. While the exact cause of SUDEP is unknown, researchers do know its highest risk is for those patients whose epilepsy started early in life and remained uncontrolled into adulthood.

Annually, more than 1 out of 1,000 people with seizures die from SUDEP; SUDEP is considered more frequent in those who have poor seizure control. Even though SUDEP is a rare complication, many in the TSC community fear having a loved one with epilepsy die because of a seizure.

Unfortunately, SUDEP is often not discussed between people with TSC, their families and their medical teams. It is not uncommon for many within our community to have never heard about SUDEP or how to take proactive measures to reduce its risks over the lifespan. Interestingly, research has shown many medical providers do not bring up SUDEP as they report not wanting to create additional stress and fear. In contrast, patient and caregiver surveys continue to report many want to know about this fatal complication even if it provokes additional stress. Many families have voiced they fear this complication, and the lack of discussion regarding the topic drives anxiety more than if providers discussed this risk with them.

The TSC Alliance is dedicated to informing families about SUDEP to ensure no one experiences this tragedy unaware and alone. Although it’s a difficult conversation, awareness about this topic can help lower SUDEP risk. In addition, families can proactively plan alternative actions to monitor more closely during times when their loved one is at high risk.

A common misconception many have regarding SUDEP is that it primarily occurs during childhood or adolescence. In fact, the risk of SUDEP is greatest during young adulthood. Years of research have helped us understand more clearly that the correlation between the age of when seizures started to how long an individual has refractory epilepsy (uncontrolled seizures) drives the major risk for SUDEP. Therefore, all efforts to reduce seizures in childhood, ensure medication adherence, develop proper sleep hygiene and elimination of drug or alcohol use are essential steps to prevent this horrific complication. Furthermore, there are more ways you can proactively help minimize the risk for SUDEP (see below). You can also find the most current information on SUDEP on the TSC Navigator.

### **Steps to reduce SUDEP**

- Control your seizures. Have regular discussions with your healthcare provider if seizures are not controlled.
- Take your medication as prescribed and try not to miss a dose. Set reminders or alerts if you have difficulty remembering. Talk to your doctor about what actions you should take if you miss a medication.
- Have a seizure action plan. Make sure to update frequently and give it to others who would need to utilize it.
- Make sure you read and understand over-the-counter medications and dietary supplements and how they may interact with your current medicines. Discuss which over-the-counter medications are safe to take or who should be avoided with your healthcare provider. Check with your pharmacist on what could interfere with your current seizure medicines. For example, medication such as Benadryl are in many over the counter medication for colds and allergies. Benadryl and other antihistamines in its class can lower your seizure threshold and should be avoided.
- Create a balanced sleep schedule and practice good sleep hygiene. This is extremely important during times of transition such as college, semi-independent, and independent living. As SUDEP often occurs at night, discuss with close family members and friends who may be of assistance.
- Avoid drugs and alcohol. These can interfere with your seizure medication or lower your seizure threshold.
- Those looking to start a family should proactively discuss with your healthcare provider prior to becoming pregnant. If you suspect you might be pregnant, please notify your health care provider as soon as possible to confirm pregnancy. Shifting hormones during this time can lead to an increase in seizures. Some medication may need to be changed and avoided during pregnancy. Your neurologist will be able to discuss these options with you.

## Section Fifteen Appendix

### SUDEP resources

| Organization  | Description  | Website  |
|---|--|--|
| <b>SUDEP Program</b>  | The Epilepsy Foundation's SUDEP program works to prevent SUDEP, and support people bereaved by SUDEP and other epilepsy-related deaths.  | <a href="http://www.epilepsy.com">www.epilepsy.com</a>   |
| <b>Centers for Disease Control and Prevention (CDC)</b>     | <p>CDC is the nation's leading science-based, data-driven, service organization that protects the public's health.</p> <p><i>Notable Resource:</i><br/> <i>SUDEP Information for Parents of Children with Epilepsy</i></p>   | <a href="http://www.cdc.gov">www.cdc.gov</a>   |
| <b>TSC Alliance</b>   | SUDEP Awareness  | <a href="http://www.tscalliance.org">www.tscalliance.org</a>   |
| <b>PAME: Partners Against Against Mortality in Epilepsy</b> | PAME's mission is to convene, educate and inspire all stakeholders - from the bereaved to those living with epilepsy, to health care professionals, advocates, clinical and basic scientists, and death investigators - to promote understanding and drive prevention of epilepsy-related mortality.   | <a href="http://www.pameonline.org/">www.pameonline.org/</a>   |
| <b>Cure Epilepsy</b>  | <p>Mission is to find a cure for epilepsy, by promoting and funding patient-focused research.</p> <p><i>Notable Resource:</i><br/> <i>SUDEP</i></p>  | <a href="http://www.cureepilepsy.org">www.cureepilepsy.org</a>   |
| <b>University of Maryland</b>                               | <p>Makes arrangements for this type of precious gift on behalf of the TSC Alliance.</p> <p>Successful donation rests on swift and thorough communication between family members, healthcare professionals, and the MBTB. Although the MBTB will make every effort to retrieve tissue in an emergency, tissue recovery may be impossible if there is no advance notice. Please click here for instructions on how to register in advance for postmortem donation. There is no cost to the family to donate.</p> | <p>Individuals living anywhere in the United States who wish to donate whole brain tissue after death should contact MBTB at <a href="mailto:btbumab@som.umaryland.edu">btbumab@som.umaryland.edu</a>.</p> <p>If death is imminent and you would like to donate brain tissue, please call the MBTB as soon as possible at 1-800-847-1539</p> |
| <b>Child Neurology</b>                                      | <p>SUDEP Resources and Tools from the Child Neurology Foundation</p> <p><i>Notable Resource:</i><br/> <i>SUDEP</i></p>   | <a href="http://www.childneurologyfoundation.org">www.childneurologyfoundation.org</a>   |







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