

Navigating the adult years of TSC

An information guide to navigate
tuberous sclerosis complex (TSC)
in the adult years.



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 our online TSC Navigator tool (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)

Some information in this guide has been introduced in the previous guides. This adult guide is for individuals and/or their caregivers continuing their TSC journey and those who are newly diagnosed.

PDF versions of each guide with clickable links are available at www.tscalliance.org.

Contents

Section One: I have just been diagnosed, now what?	3
Where do I start?	3
What are my options?.....	3
Where do I go?.....	3
How do I get connected?.....	4
Section One Appendix.....	5
Section Two: Navigating the medical journey.....	8
Understanding the diagnosis	8
Clinical manifestations	8
2021 TSC diagnostic criteria updates	9
TSC genetics.....	9
Clinical management and your healthcare team	9
Staying organized.....	10
TSC over time	10
Neurological manifestations.....	10
Kidney manifestations	10
End-stage renal disease.....	11
Lung manifestations.....	11
Heart manifestations	11
Skin, eyes and teeth manifestations	11
Other manifestations	12
Medical challenges.....	12
Pharmacy coverage	12
Prior authorization issues.....	12
Insurance barriers	13
Tips to mitigate issues.....	13
Medication assistance	14
Health insurance and medical benefits	14
Clinical trials and research opportunities	15
What is a clinical trial?	15
Why participate in a clinical trial?	15
Who can participate in a clinical trial?.....	15
What are the benefits and risks of participating in a clinical trial?.....	15
Where do I find a clinical trial for TSC?.....	15
Where can I get more information about clinical trials?	15
Section Two Appendix	16
Section Three: TSC-associated neuropsychiatric disorders (TAND) and mental health.....	33
What is TAND?	33
Assessment of cognitive and behavioral health issues	33
Therapies and services	33
Mental health and TSC.....	34
Crisis intervention plan	34
Section Three Appendix.....	35
Section Four: Legal rights under disability law	36
Americans with Disability Act (ADA).....	36
Legal assistance	36
Family and Medical Leave Act (FMLA)	36
Air Carrier Access Act.....	36
Telecommunications Act.....	36
Section Four Appendix	37

Section Five: Employment, vocation and housing	38
Employment rights.....	38
Vocational/rehabilitation programs, jobs and employment.....	38
Federal employment and support programs.....	38
How do I find a job?.....	39
Residential programs.....	39
Public housing.....	39
How to qualify.....	39
Section 811.....	39
Section Five Appendix.....	40
Section Six: Life and financial planning	41
Financial and estate planning.....	41
Guardianships.....	41
Alternatives to guardianships	42
Section Six Appendix	43
Section Seven: Assistive technology (tools and devices)	44
What is assistive technology?	44
What is the purpose/benefit of assistive technology?.....	44
How do you fund these services?.....	45
Other tools and devices.....	45
Section Seven Appendix	46
Section Eight: Self care and family dynamics	47
Family planning	47
Reproductive decision making and TSC.....	47
Prenatal screening	47
Pregnancy prevention.....	47
Self care and mental health.....	48
Adult self care and mental health.....	48
LGBTQIA+ community and TSC.....	49
Semi-independent adult mental health.....	49
Caregiver mental health	49
Signs and symptoms of caregiver burnout.....	50
Aging caregiver	51
Respite care	51
Section Eight Appendix.....	52
Section Nine: Difficult conversations and end of life issues	63
Continuum of grief.....	63
SUDEP.....	64
Steps to reduce SUDEP.....	64
Advanced decision making.....	65
Advanced decision making planning considerations	65
Acute decision making considerations	67
Hospice care	69
What is hospice?	69
How does hospice care work?.....	69
How do I find hospice care?.....	69
Who pays for hospice care?.....	69
Grief and bereavement	70
Blood and tissue donation	70
Natural History Database	70
Blood samples	70
Tissue samples.....	71
Post-mortem brain donation.....	71
Section Nine Appendix.....	72

Section One: I have just been diagnosed, now what?

Where do I start?

Receiving a diagnosis of a rare genetic disease as an adult can be confusing and overwhelming. Depending on the findings, it is important to have ongoing conversations with your healthcare team about what to expect. In adulthood this can be by an incidental finding (meaning it was found by looking for something else) or an abrupt finding (such as a renal hemorrhage), which can be even more overwhelming to process. Regardless of the presenting feature, once a diagnosis has been made or suspected it is important for you to undergo a thorough evaluation.

Key factors in evaluating TSC are outlined in the 2021 Updated TSC Diagnostic Criteria (see Section Two). It is important to establish care with a clinician who can provide proper surveillance and treatment recommendations across the lifespan. TSC is unique, and every journey starts differently so it's okay if you are feeling overwhelmed and not sure what to expect. This guide will help you walk through some of these questions and provide you with the resources to proactively empower your next steps. You can refer to the New Diagnosis Flowchart in the appendix of this section to see how your journey might start.

What are my options?

Treatment options directly depend on the symptom or the organ system causing an issue. Adulthood diagnosis of TSC typically presents much differently than childhood, therefore treatment management is usually associated with a chronic issue you may or may not have had an explanation of prior to the diagnosis. Often, people describe mixed emotions of relief and concern during this process. Regardless of how your journey started, maintaining an open line of communication with your healthcare team is essential.

Treatment options in this life stage typically focus on kidney, lung and psychological (anxiety, depression, memory) management. Even though it is uncommon to develop seizures during this life stage, many treatment options are available for TSC-related seizures. Other treatment options focus on high blood pressure (hypertension) or proteinuria (protein in urine) management. This is common in those with renal involvement. If you have 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. Many options exist for blood pressure and proteinuria management.

Lung involvement is also another adult-specific diagnosis. The associated disease, lymphangioleiomyomatosis (LAM), typically presents in post-pubescent women (it has only rarely been observed in men).

Advances in research continue to deliver new and improved therapeutic options. There are now five (5) FDA-approved treatments in the United States available for some aspects of the disease and related conditions. These approved options include: 1. Afinitor® (everolimus), 2. Rapamune® (sirolimus), 3. Sabril® and Vigadrone® (vigabatrin), 4. Epidiolex® (cannabidiol) and 5. Hyftor® (sirolimus topical gel).

After allowing time to process the initial diagnosis of TSC, you can start navigating options with your treatment goals with your healthcare team. Understanding all your treatment options at every stage of your journey is essential. Depending on your diagnosis, a clinical trial may be an option. Learn more about clinical trials in Section Two.

Where do I go?

It is common for medical providers to be unfamiliar with TSC, including neurologists and primary care providers (PCPs). It is crucial to have a local healthcare team involved in your care. Finding a good fit is essential as these healthcare team members will be your experts and, along with you, your biggest advocates as you start your TSC journey. Connecting with a local or out-of-state TSC clinic to help guide clinical treatment options is appropriate. You can locate a TSC clinic by visiting www.tscalliance.org/tscclinics.

If you do not have access to a local TSC clinic, you can also make an appointment with an adult or pediatric neurologist experienced in epilepsy. Many of our TSC experts can offer provider-to-provider recommendations, and our TSC Support Navigators can assist with this process. To find more information about the TSC Support Navigators, please visit the TSC Navigator on www.tscalliance.org.

Insurance coverage can also be a factor in ensuring you do not experience delayed intervention and care. It is important to consider if out-of-state healthcare is right for you. For those considering traveling across state lines, please go to the insurance barriers page in the TSC Navigator's Medical Challenges section to learn more on how to proactively prevent issues from occurring.

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, so our TSC Support Navigators are available to help you at any time.

This journey can feel lonely at times but connecting with another individual who has navigated these steps before you can make all the difference. To gain more insight on self care and family dynamics, please go to Section Eight to learn more. In addition, there are plenty of ways you can connect with the TSC community. 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 TSC Alliance initiatives, videos, town halls, 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
- Facebook: facebook.com/tscalliance
- Instagram: instagram.com/tscalliance
- YouTube: youtube.com/tscalliance

Facebook options

Connect with other members of the TSC community in the private TSC Alliance Tuberous 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.

Volunteer opportunities

Volunteering offers an opportunity to connect and engage with other individuals 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 www.tscalliance.org for more information.

Peer-to-peer support

Adult Regional Coordinators in eight regions across the United States serve as a connection for other adults diagnosed with TSC. They are knowledgeable about resources, events, support meetings, conferences and any new research on TSC that pertains to adults with TSC. To connect with an adult regional coordinator, go to www.tscalliance.org/individuals-families/adults/.

Adult Topic Calls

These monthly calls are for independent or semi-independent adults with TSC. Calls range from open forums to specific topics where adults can discuss challenges and success in a safe environment while building long lasting friendships. You can find more information on dates and times for the Adult Topic Calls on the TSC Alliance website.

TSC Waves

TSC Waves is a quarterly e-newsletter for adults with TSC from the TSC Alliance featuring articles written by adults with TSC, profiles on our Adult Regional Coordinators, a schedule of upcoming events and educational sessions relevant to adults. *TSC Waves* also includes resources from the TSC Alliance to help adults cope with the myriad of manifestations and challenges they face living with tuberous sclerosis complex. You can sign up for this e-newsletter at www.tscalliance.org/tscwaves.

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
<p>TSC Navigator</p>	<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>	<p>www.tscalliance.org/tscnavigator</p>
<p>TSC Now</p>	<p>A podcast series 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>	<p>www.tscalliance.org/tscnow</p>
<p>TSC Matters</p>	<p>A bimonthly community e-newsletter from the TSC Alliance highlighting TSC research updates, TSC clinical trials, upcoming TSC Alliance events and more.</p>	<p>www.tscalliance.org/tscmatters</p>
<p>TSC Academy</p>	<p>An online educational platform created for the TSC community. TSC Academy 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>	<p>www.tscacademy.org</p>
<p>Webinars and Videos</p>	<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>	<p>www.tscalliance.org/webinarsandvideos</p>

Other notable resources

Organization	Description	Website
The Arc	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.	www.thearc.org
Understood for All, Inc.	Online resource to help those who learn and think differently discover their potential, take control, find community, and stay positive along each life stage.	www.understood.org
Epilepsy Foundation	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.	www.epilepsy.com
PACER Center, Inc.	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.	www.pacer.org
Nation Patient Advocate Foundation (NPAF)	Provides educational resources to help patients advocate for themselves and make informed, personalized health care decisions.	www.npaf.org
Patient Advocate Foundation (PAF)	Provides professional case management services to patients with chronic, life threatening and debilitating diseases.	www.patientadvocate.org



*The TSC Alliance offers a wide range of resources for adults with TSC and their caregivers.
Find out more at www.tscalliance.org/adults.*

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 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.

Another way to get informed is with TSC Academy, an online educational platform created for the tuberous sclerosis complex (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 along with federal, state and local governments and other stakeholders work together to find a cure; and how you play a role.

When you are ready, learning about common TSC manifestations during adulthood is a helpful guide to proactively give insight on what to look out for during the next phases of the journey. Learn more about signs and symptoms of TSC on the TSC Navigator under Tips from Adults.

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 assist clinicians to identify major and minor features that are considered “hallmark” findings of TSC.

In 2021, the clinical diagnostic was updated and is reflected below. Not all features of TSC are listed, but not all ailments are associated with TSC, so it's important to have ongoing discussions with your TSC expert provider so proper treatment recommendations can be initiated or referral to other specialists is not delayed.

TSC diagnostic criteria

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

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

Possible TSC: Either 1 major feature or >2 minor features.

*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.

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 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 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 figure out 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 visit TSC Academy.

Clinical management and your healthcare team

You do not have to be an expert in TSC to navigate your care. You can empower yourself by clearly advocating for your understanding of TSC and what you need to better understand, assessing risk and benefit of treatment, determining 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 yourself under the care of many different medical specialists. Most of these specialties are separated between adult and pediatric patients. Two medical specialties are crucial to helping you manage and coordinate all these specialists: your internist (adults) or pediatrician (children). 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 during 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 a clinical trial. The Bcureful Travel Fund at the TSC Alliance underwrites the TSC Bcureful Travel and Lodging Patient Assistance Program, which is administered by the National Organization for Rare Disorders (NORD). The TSC Bcureful Travel and Lodging Patient Assistant Program 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.

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 that 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.

As this is the first phase of your journey, there are many laboratory and diagnostic tests that will be mentioned along the way. Having an understanding 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 are critical with this diagnosis. Building a solid foundation now will make a huge difference during your journey. Learn more about Medication and Testing in the appendix of this section.

Trying to stay organized when you are an individual with TSC or a caregiver to an individual requires various appointments and 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. For downloadable versions, go to the TSC Navigator's Resources section.

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 guides 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 the 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 Four. 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.

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 – 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 ideal 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 your 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 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 – 3 years or more frequent pending your unique situation. Proactively understanding symptoms that you should report to your clinical team would include connective tissue 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 has 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 your fingernails or toenails) can also increase during adulthood. If those become problematic or bothersome, surgical removal can be completed by a dermatologist.

At this time, 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 GI 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.

Medical challenges

Pharmacy coverage

Prescription drug plans are part of all commercial government insurance plans in the United States; however, each plan is different per corporation/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 list which medications will be covered under your plan (called a formulary), but what category or "tier" each of these medications are listed into. Tiers represent a cost level. There are 4 tiers to a formulary list with tier 1 being the lowest cost and tier 4 being the highest cost. Tiers 1 to 3 usually require some co-payment, 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-name drugs you can use per month (this includes 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 copayment 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%-100%) of medication costs. This is why it is very important to understand your deductible plan, since progress toward "meeting your deductible" (i.e., the amount of money you spend each year) resets annually.

Many people utilize copay assistance cards so medications can be affordable; however, under the current co-pay accumulator programs, this 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) located 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 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 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.

Prior authorization issues

Medications prescribed for TSC-related manifestations often require a prior authorization (PA). It is important to know which medications will require this 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 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 where individual lives) 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 of 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 there are multiple insurances 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 the state's Medicaid program.
 - o Contact the out-of-state clinic prior to the referral being placed to see 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 that will be seeing you or your loved one.
 - 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 all together 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 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 a prior authorization (PA). 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 (Afinitor®), but your plan will only cover generic (everolimus). If this substitution is allowed, then call your prescribing provider's office to let them know as soon as possible. Never assume your provider or office staff 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 (help!): 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 to 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 still in need of financial assistance. While these programs often limit the total amount an individual can utilize per year, it is helpful to know which options are available. Co-pay assistance programs are typically for medications that are brand name with no generic equivalent available. Often, your prescribing providers or specialty pharmacists 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. We do advocate proactively seeking opportunities to help our community to utilize 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, they 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 Support Navigators for assistance.

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.

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 or your local Department of Human Services.

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 www.tscalliance.org/clinicaltrials.

Where can I get more general information about clinical trials?

The U.S. National Library of Medicine maintains 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 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 Two Appendix

Organization	Description	Website
Medicaid.gov	<i>Notable Sections: Beneficiary Resources</i>	www.medicaid.gov
Centers for Medicare and Medicaid Services (CMS)	Provides an extensive list of state-based resources on its website.	www.medicare.gov/care-compare/
Kaiser Family Foundation (KFF)	Is dedicated to filling the need for trusted information on national health issues.	www.kff.org/statedata/
Medication Assistance Tool (MAT)	MAT is a free-to-use search engine that focuses its searches on patient assistance resources available to eligible patients.	www.mat.org
Needy Meds <i>Languages Available: English, Spanish</i>	NeedyMeds connects people to programs that will help them afford their medications and other healthcare costs	www.needymeds.org
RxAssist	RxAssist offers a database of patient assistance programs, and offers a prescription drug savings/discount card at no cost	www.rxassist.org
Step Therapy	It is a policy developed by health insurers that is intended to control costs.	www.steptherapy.com
Prescription Process	A non-profit national network of physicians who advocate for patient access to approved therapies and appropriate clinical care.	www.prescriptionprocess.com
FDA Drug Shortages	Current and Resolved Drug Shortages and Discontinuations Reported to FDA	www.accessdata.fda.gov/scripts/drugshortages/
GoodRx	Regardless of an individual's income and insurance status, GoodRX helps people find the right care for the right price.	www.goodrx.com
Total Care Rx	A comprehensive pharmacy solution bringing together specialty pharmacies, technicians, customer service and delivery staff to deliver beyond the prescription.	www.totalcarerx.com
Chemistry Rx	A compounding pharmacy dedicated to providing customized medication to patients, providers, and other organizations across the entire healthcare system.	www.chemistryrx.com
Kidney Foundation	The Kidney Foundation is a lifeline for all people affected by kidney disease.	www.kidney.org/
PKD Foundation	Solely dedicated to finding treatments and a cure for polycystic kidney disease (PKD). The fund research, education, advocacy, support and awareness on both the national and local level.	www.pkdcure.org/
LAM Foundation	The LAM Foundation seeks safe and effective treatments, and ultimately a cure, for LAM through advocacy and the funding of research.	www.thelamfoundation.org



2021 TUBEROUS SCLEROSIS COMPLEX

DIAGNOSTIC CRITERIA, SURVEILLANCE AND MANAGEMENT RECOMMENDATIONS

Diagnostic criteria

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

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











Possible TSC: Either 1 major feature or ≥ 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	
GENETICS	<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>	
BRAIN	<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>	
TAND	<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>	
KIDNEY	<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>	
LUNG	<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>	 
SKIN	<p>Perform a detailed clinical dermatologic inspection/exam.</p>	
TEETH	<p>Perform a detailed clinical dental inspection/exam.</p>	
HEART	<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>	
EYE	<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 (tandconsortium.org/checklists/). 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>RENAL</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>PULMONARY</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 < 70% predicted), physiological evidence of substantial disease burden (abnormal DLCO (<80% or less than lower limit of normal [when available]), air trapping (RV > 120%), resting or exercise-induced oxygen desaturation, rapid decline (rate of decline in FEV1 > 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>SKIN</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>TEETH</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>HEART</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>EYE</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>OTHER:</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>	



8737 Colesville Road, Suite 400 | Silver Spring, MD 20910
800-225-6872 | info@tscalliance.org | tscalliance.org

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										
Internist												
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: _____ Name: _____ Type (phone, email): _____

Notes: _____

Date: _____ Name: _____ Type (phone, email): _____

Notes: _____

Date: _____ Name: _____ Type (phone, email): _____

Notes: _____

Date: _____ Name: _____ Type (phone, email): _____

Notes: _____



NORD[®]
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.[®]

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.

©2021 NORD. All rights reserved. NORD, its icon, RareCare and tagline are registered trademarks of The National Organization for Rare Disorders. NORD is a registered 501(c)(3) organization. NORD does not recommend or endorse any particular medical treatment but encourages patients to seek the advice of their clinicians. Donations to NORD for this and other programs may be made by contacting NORD at rarediseases.org. NRD-2131



rarediseases.org

Section Three: TSC-associated neuropsychiatric disorders (TAND)

What is TAND?

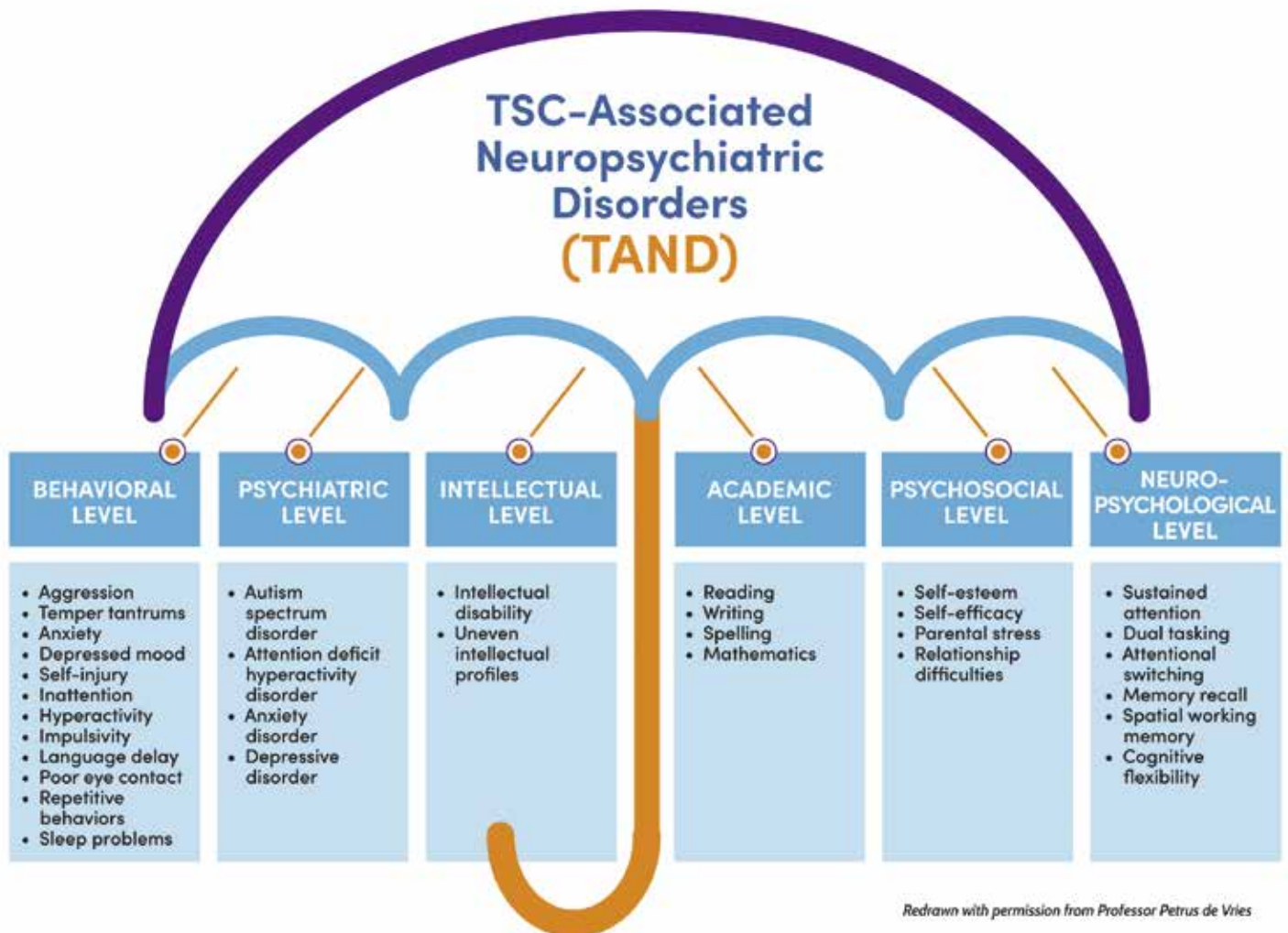
Even though TAND affects 90% of those with TSC over their lifetime, everyone experiences unique symptoms, so interventions may be different from individual to individual. In the Behavioral Issues and TAND guide on the TSC Navigator, you will find detailed information regarding each of the impacts of the six major areas.

Other helpful resources can be found on the TANDem website 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 BCBA 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.



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 Videos 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, portable treatment records which include what behaviors you would call 911 over or to seek emergency support. They also provide a 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.

Section Three Appendix

Organization	Description	Website
TANDem	TANDem is an international multi-disciplinary mobile-health project to empower families and individuals who live with Tuberous Sclerosis Complex (TSC) around the world.	www.tandconsortium.org
National BACB	Search engine for finding certified therapists in your area.	www.bacb.com
YAI Seeing Beyond Disability	YAI is a network of affiliate agencies offer children and adults with intellectual and developmental disabilities a comprehensive range of services.	www.yai.org
Devereux Advanced Behavioral Health	Devereux Advanced Behavioral Health changes lives – by unlocking and nurturing human potential for people living with emotional, behavioral, or cognitive differences.	www.devereux.org
NeuroRestorative	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.	www.neurorestorative.com
National Alliance on Mental Illness (NAMI)	<p>Provides advocacy, education, support and public awareness so that all individuals and families affected by mental illness can build a better life.</p> <p><i>Notable Article: Being Prepared for a Crisis</i></p>	www.nami.org

Section Four: Legal rights under disability law

Americans with Disability Act (ADA)

The ADA prohibits discrimination against individuals with disabilities in employment, housing, education, and access to public services (transportation, housing, etc.).

How does the ADA define “disability”? To be protected by the ADA, a person must have one of the following:

1. A physical or mental impairment that substantially limits one or major life activities of the individual.
2. A record of such impairment.
3. A perception by others as having such an impairment.

The ADA further requires that reasonable accommodation be made to provide individuals with disabilities equal opportunities. Federal agencies and departments charged with enforcing the ADA include the Equal Employment Opportunity Commission (EEOC) and the Department of Justice. Specific titles of the ADA (I-IV) address specific rights regarding employment, state and local government activities; public transportation; public accommodations; and telecommunication relay services. States may pass disability statutes so long as they are consistent with the ADA.

Legal assistance

The TSC Alliance acknowledges legal assistance is sometimes needed to help with certain barriers or challenges that are unique to your situation for appropriate resolution. As an organization, we cannot endorse an individual or organization. The information in this section simply serves as a reference point and a place to start to access resources.

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 Four Appendix

Organization	Description	Website
Social Security Administration	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.	www.ssa.gov/disability/
U.S. Department of Justice Civil Rights Division	A guide to disability rights laws.	www.ada.gov/cguide.htm
Learning Disabilities Association of America (LDA)	LDA's mission is to create opportunities for success for all individuals affected by learning disabilities through support, education, and advocacy.	www.lidaamerica.org
The Council of Parent Attorneys and Advocates (COPAA)	An organization of attorneys, advocates and parents established to improve the quality of legal assistance for parents of children with disabilities.	www.copaa.org
US Department of Labor (DOL) Office of Disability Employment Policy (ODEP)	Provides fact sheets regarding disability issues, discrimination, and legal rights	www.dol.gov/odep

Section Five: Employment, vocation and housing

Employment rights

As covered in Section Five, the 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 the person can effectively perform their job, such as providing wheelchair accessibility. For more information, please visit 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 co-workers 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 a number of services for people with disabilities who are 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 nonprofit 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 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.

Project Employ is a joint initiative of the Office of Disability Employment Policy (ODEP) and the Society for Human Resource Management (SHRM) 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 the 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 is 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 <https://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.

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

How to qualify

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 Five Appendix

Organization	Description	Website
Job Accommodation Network (JAN)	A source for free, expert, and confidential guidance on job accommodations and disability employment matters.	www.askjan.org
HUD	U.S Department of Housing and Urban Development <i>Notable Section: Disability Overview</i>	www.hud.gov
National Council of State Housing Agencies (HCSHA)	Assist the Housing Finance Agencies' efforts to provide affordable housing to those who need it <i>Notable Article: Special Needs Housing</i>	www.ncsha.org
Ability Jobs	Works with companies, government and nonprofit agencies to employ people with disabilities.	www.abilityjobs.com
Equal Opportunity Publications. Inc. (EOP)	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
Housing Center for People with Disabilities	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. <i>Notable Sections: Housing News Headlines, Opening Doors, HUD Programs, and Federal Housing Policy and Legislative Updates.</i>	www.tacinc.org
Mainstream	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.	www.mainstreaminc.net
Independent Living	The NCIL provides resources regarding independent living and a national directory of Centers for Independent Living.	www.ncil.org
Bender Consulting	Consulting firm that specializes in recruiting and hiring people with disabilities for full-time, competitive employment opportunities.	www.benderconsult.com

Section Six: 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. 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 of 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 of 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 that 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. A **Guardian of the Person** is responsible for monitoring the care of the ward.
- b. 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.
- c. 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.
- d. 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 of the 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 is capable of choosing another person to handle his/her 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 Six Appendix

Organization	Description	Website
Special Needs Alliance (SNA)	A national organization of attorneys committed to the practice of disability and public benefits law. Individuals with disabilities, their families and their advisors rely on the SNA to connect them with nearby attorneys who focus their practices in the disability law area. The mission of the SNA is to maintain a professional organization of attorneys skilled in the complex areas of public entitlements, estate, trust and tax planning, and legal issues involving individuals with physical and cognitive disabilities.	www.specialneedsalliance.org
MassMutual	Committed to help people secure their future and protect their loved ones. <i>Notable Section: Financial Wellness and Special Needs</i>	www.massmutual.com
National Guardianship	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.	www.guardianship.org
Future Planning	This resource list from The Arc includes government agencies, publications regarding estate planning, guardianship financial and future planning resources by state. <i>Notable Article: Future Planning</i>	www.thearc.org
National Law Review	<i>Notable Article: Understanding Special Needs Trusts</i>	www.natlawreview.com
Veterans Affairs	<i>Notable Section: Family and Caregiver Health Benefits</i>	www.va.gov

Section Seven: 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.

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 people with disabilities can use either to:

1. assist them in learning
2. make the environment more accessible
3. enable them to compete in the workplace
4. 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:

1. Access to computerized information on the use of assistive technology
2. Develop demonstration centers where people with disabilities can try different pieces of equipment
3. Consultants to help individuals make informed decisions about equipment
4. Referral services
5. 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. 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."

Other tools and devices

While assistive technology encompasses many tools and devices that can support enhanced independence and participation in education and other aspects of life, there are also other resources outside of these categories to consider, such as service animals. Under the ADA, a service animal is defined as a dog that has been individually trained to do work with or perform tasks for an individual with a disability. A service animal must be trained to take specific action when needed to help someone with their disability or aspects of their disability. The ADA also sets the rules for how service animals can access public and private spaces, such as restaurants, hospitals, schools, hotels and housing.

Common examples of service animal work can include alerting for seizures, guiding blind individuals or providing other assistance and day-to-day support. Service animals can provide specific support, such as alerting for seizures, but may also have additional benefits in supporting the quality of life, independence and social interaction of a disabled individual.



Section Seven Appendix

Assistive tools and devices

Organization	Description	Website
Seizure Tracker	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.	www.seizuretracker.com
Danny Did Foundation	A resource for families who are impacted by seizures <i>Notable Section:</i> <i>Devices + Technology</i>	www.dannydid.org
Epilepsy Foundation	To lead the fight to overcome the challenges of living with epilepsy and to accelerate therapies to stop seizures, find cures, and save lives. <i>Notable Section:</i> <i>Epilepsy Device Wiki</i>	www.epilepsy.com
Charlie Foundation	Dedicated to advancing the awareness of ketogenic therapeutics to help with seizures.	www.charlifoundation.org
Chelsea Hutchison	A non-profit to provide help and support to individuals, particularly children and young adults, who have epilepsy. <i>Notable Section:</i> <i>Helpful Links: Monitoring Devices</i>	www.chelseahutchisonfoundation.org

Service dogs

Organization	Description	Website
Domesti-PUPS	A community organization improving the quality of life for individuals with special needs through the assistance of animals and raising awareness through education.	www.domesti-pups.org
Domino Service Dogs	Assists individuals with disabilities to train their own service dogs, using reward-based training.	www.dominoservicedogs.com
Little Angels Service Dogs	A nonprofit partnering service dogs with the disabled, helping to empower and provide independents for the recipients.	www.littleangelsservicedogs.org
New Hope Assistance Dogs, Inc.	Dedicated to enhancing the lives of individuals with disabilities and educating the public with service dogs.	www.newhopedogs.net
4 Paws for Ability	A nonprofit dedicated to enriching the lives of individuals with disabilities by pairing them with life changing service dogs.	www.4pawsforability.org

Section Eight: Self care and family dynamics

This section blurs between the needs of an independent adult, semi-adult, dependent adult to the aging caregiver; however, while the importance of establishing healthy boundaries, self care and the unique challenges alter slightly, the foundation of these topics is the same. We have categorized each of these topics into their own categories; therefore, if it does not align to your current situation you can move to the categories that do.

Family planning

Reproductive decision making and TSC

Deciding to start a family or grow an existing family is a life-changing decision full of excitement and hope. However, if your family medical history contains a diagnosis of TSC, this excitement and hope can give way to fear and anxiety. Fortunately, geneticists can support families in reproductive decision making. The geneticist, along with a team of genetic professionals, tries to meet three goals with the family: diagnosis, prognosis and recurrence risk.

For the purposes of reproductive decisions, when the diagnosis of TSC is already known within a parent or family member, recurrence risk is often the most crucial information a family is seeking. A recurrence risk means the risk (or odds) that a subsequent child will be affected with the same condition. In the case of diagnosing TSC, the genetic professional's job is to convey the genetic facts about TSC. TSC is a genetic disease caused by a change in the TSC1 or TSC2 gene that is causing it to no longer work. The non-working gene can then be passed on to future generations. However, it is important to remember approximately two-thirds of the time when a child is diagnosed with TSC, neither parent has TSC. If the change in TSC1 or TSC2 that causes TSC in a family member can be detected and identified, further testing can be performed on other family members or used for prenatal testing.

Prenatal screening

Prenatal genetic testing is a common method many women seek to learn whether their babies have certain genetic disorders. There are two types of prenatal tests: prenatal screening tests and prenatal diagnostic tests. Screening tests focus on your risk of having a baby with certain disorders, such as carrier screening and prenatal genetic screenings. Carrier tests can be completed before or during pregnancy.

Prenatal genetic screenings (such as an ultrasound or by testing the pregnant woman's blood) focus on finding unexpected changes to major organs and blood components. Women with a known genetic disorder, such as TSC, can benefit from prenatal diagnostic tests. This type of testing focuses on whether the fetus has inherited a certain disorder. Testing is performed on fetal cells, placenta, amniocentesis or chorionic villus sampling.

It is important to understand a positive screening test does not confirm a disorder – rather, it means your baby has a higher risk of a disorder. Follow-up diagnostic testing is required to confirm a screening result. We strongly recommend discussing options with your healthcare team, which at this stage might include a genetic counselor, to consider the meaning of your results and options moving forward.

You can learn more by referencing Reproductive Decision Making and TSC in the appendix of this section. If you have TSC and are considering pregnancy or are pregnant, you can find more information on how to navigate an in utero (during pregnancy) pregnancy. It is also important to consider talking to your medical team prior to conceiving as there are certain manifestations (such as seizures, renal lesions and/or LAM) your medical team will want to monitor more closely, in addition to altering any medications that are not safe during pregnancy. It is also common for women to experience more seizures during this time even if your seizures have been controlled for many years. If you are currently pregnant and have not notified your medical team, you should do so as soon as possible.

Pregnancy prevention

For those who were assigned female at birth and who wish not to get pregnant for whatever reason at this time, it is medically recommended to avoid estrogen therapy and focus on other available options. As this list is consistently changing, the most important factors are to discuss with your local medical provider what options you should avoid so he/she can outline options that are best for you. As with all areas of medicine, advancement with birth control options continue to improve. It is essential to align to an option that is realistic, sustainable and consistent if you or your loved one with TSC is sexually active.

Other TSC manifestations that should be considered with your provider as this discussion takes place includes whether you or your loved one is affected by high blood pressure, has a history of cancers (specifically breast cancer) and or has had a blood clot. Most healthcare plans cover all the available options including contraceptive pills, injections, intra-uterine devices and implants. You can look on your pharmacy drug list under “birth control” or “reproductive contraceptives” to see what your plan covers.

For those with TSC who were assigned male at birth, you still have a 50% chance of passing TSC to your offspring; therefore, contraceptive methods are strongly recommended. Despite many medical advancements in healthcare, men’s options for quality and functional contraceptive methods are still limited to condoms and vasectomies. While both are effective means to prevent pregnancy, discussing long-term goals with your local medical provider is recommended. Condoms also provide excellent protection against sexually transmitted infections (STIs) such as HIV, chlamydia, gonorrhea and syphilis. A vasectomy is a medical procedure that is considered to permanently prevent pregnancy, known as male sterilization. This is considered invasive, and you will have some recovery time; however, you do not go under anesthesia for this procedure in most situations. In addition, the cost of a vasectomy is covered under your insurance including for those who have Medicaid.

For the aging caregiver, it is important to remember that if your loved one with TSC is semi-independent or dependent, he or she may still have sexual desires. This issue may pertain to some of these adults more than others, and only you as a caregiver will know your loved one’s true cognitive abilities. For semi-independent or dependent adults who go to camps, day camps, schools or other group settings, it is important to have discussions with them about appropriate and inappropriate sexual relationships. As much as these settings try to prevent these opportunities, sexual situations do still occur. It is best to not avoid this topic, even though it might be uncomfortable. The other issue, despite being an unfathomable reality, is that many with special needs are targets of abuse and sexual assault. Depending on your loved one’s cognitive abilities and level of understanding, it is important to have discussions about appropriate and inappropriate situations in a way your loved one can easily comprehend.

Role playing basic scenarios may also be very helpful to those who struggle with social cues. Regardless of your love one’s gender, if you suspect he/she has been abused or sexually assaulted, you need to immediately notify your local police authorities and the primary care provider. You can also reach out to the National Sexual Assault Hotline at 1-800-656-4673.

Self care and mental health

Adult self care and mental health

Being diagnosed with TSC can cause significant challenges within your adult life regardless of when you first received your diagnosis. From the stress of learning a new diagnosis to potentially processing the emotions of an offspring with this diagnosis to learning how to navigate the medical complexity of routine follow-up surveillance and management in the adult healthcare sector, all while maintaining other responsibilities, can be devastating and overwhelming. 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 you’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 in adulthood, even if you are full-time employed and/or have completed graduate-level studies. The emotional challenges can be just as debilitating as physical challenges and significantly trigger ineffective coping mechanisms, stress, and anxiety.

Another common challenge many independent adults with TSC face focuses on dating, relationships and social life. From navigating discussions of reproductive choices to feelings of resentment from a significant other for “giving” this diagnosis to an offspring can create many challenges. Regardless of what your story looks like, it is important to know you are not alone, and you are not the first to have the thoughts and feelings about all the emotions that come along with this journey.

Later in this section you will learn about the signs and symptoms of caregiver burnout, which can also be applied to you as an individual living with TSC. This information will help gauge your level of stress and identify helpful resources to proactively take charge in areas you do have control over. In addition, being part of the adults with TSC community can greatly impact your emotional wellbeing by connecting with other adults traveling the same road.

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.

Those interested in gender-affirming hormone therapy (GAHT) may need to take extra precautions to ensure safety since some hormones can interfere with seizure medications and potentially TSC manifestations. At this time, we do not have any current research to guide long-term risks that would be specific to TSC and GAHT. Therefore, we strongly encourage you to work closely with a healthcare team who provides gender affirming care in collaboration with your TSC experts.

The GLMA Provider Directory is the recommended resource to locate a LGBTQI+ friendly provider to help manage your overall healthcare needs. Another important step is making sure your seizure action plan is up to date if you consider or undergo GAHT. 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 on navigating your journey.

Semi-independent adult mental health

Those with TSC who are considered semi-independent adults (SIAs) also face unique challenges during this life stage. As many of these adults have transitioned into a day program, semi-independent living or have family members as their main source of care, the mental health needs of the SIA have also changed. Although the needs of those who fall into this category greatly vary, their mental health struggles are similar. This group is also at an increased risk of social manipulation, abuse and assault. It is important to ensure a health boundary of independence but also to ensure your semi-independent loved one is getting the resources and care deserved. Having discussions regarding what is appropriate and inappropriate often needs to be revisited as new people or staff come into his or her daily routine and social activities.

For those who opt for group living situations, it is important to check in with your SIA frequently. SIAs can benefit from social skills training as well as being placed into inclusion programs, such as Best Buddies, where they can continue to be included amongst their peers, even at this age in life. Depending on your loved one's ability to verbalize concerns, it is not uncommon to see changes in behavior or mood when things are not going normally. TAND manifestations can also contribute to behavioral issues and negatively affect personal well-being such as obsessive-compulsive behaviors or difficulty adjusting to different routines. Mental health resources in the United States are limited; however, ABA and BCBA therapists continue to be excellent resources to help your loved one with any behavioral or social limitations. Speech language pathologists are other great resources who can offer social skills training to those needing additional support.

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 like TSC. Western society often considers the village as solely for the loved one who needs assistance; however, the reality is the village is the support system for the primary caregiver in addition to the person with TSC.

Regardless of where you are on the TSC journey, it is important to take a moment and realize the first step in acknowledging caregiver mental health is accepting grief started upon the diagnosis of TSC. 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 as well as researching treatment options, experts, and resources for their loved ones, 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. 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 like 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 minds and bodies in an effort to provide the absolute best for their loved ones 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. The information below covers 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, so 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 get asked, “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 you can show to others not only helps you, but it also helps those in your inner circle to know how to truly help you.

Locate the Finding your Caregiver Zone guide and How to Help Me 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 the following information to your local PCP or 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.

Aging caregiver

Growing old is an inevitable part of life, but it can be especially daunting for those who care for individuals with a rare disease and/or intellectual disabilities. Life as a caregiver (e.g., cooking, managing medicines, medical appointments, therapy, etc.) can lead to decreased attention to your own personal needs, physical wellness and mental health. According to studies on aging caregivers, these significant barriers increase your risk for health problems, decrease quality of life and may cause early death. Caregivers older than 65 have a 63% higher risk of serious health issues due to prolonged emotional and physical stress.

As caregivers age, they may feel haunted by difficult questions (“I’m not going to live forever so who is going to care for my loved one when I am gone?” or, more simply, “Will my loved one be okay?”). These questions are absolutely normal, particularly when confronting mortality. Contemplating this reality as a caregiver is difficult and can awaken some all-too-familiar feelings of fear, worry, loneliness and isolation.

Your love and dedication as a caregiver to an affected individual with TSC is an inspiration and has helped improve quality of care and life for all who are affected by the disease. To help you in your journey as a caregiver, the TSC Alliance has assembled resources to help you navigate this particularly challenging part of caring for someone with TSC. The goal is to help you cope and focus on a practical, actionable care plan for your loved one, while also being vigilant regarding your own personal health and how to identify and mitigate stress brought on by your dedication to care. Please reference the Aging Caregiver Checklist in the appendix of this section to help guide your thoughts.

Respite care

For families of an adult child/loved one with TSC, respite care is a support service that may be required from time to time. Several types of respite care exist, 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 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’s ABCs of Respite: A Consumer Guide for Family Caregivers.

Please note: Respite care and services vary by state and region.

Section Eight Appendix

Family planning resources

Organization	Description	Website
Resolve – The National Infertility Association	Provides education, advocacy, and support for people facing infertility, multiple miscarriages or having a difficult time with conception.	www.resolve.org
Society For Assisted Reproductive Technology (SART)	SART is the primary organization of professionals dedicated to the practice of assisted reproductive technologies (ART) in the United States. The mission of the organization is to set and help maintain the standards for ART to better serve its members and patients.	www.sart.org
Adoption Resources	The primary purpose of Adoption Resources is to serve the best interests of children, so each child will be raised in a permanent and loving family. Adoption Resources strives to provide services that protect the dignity of children, birth parents, adoptive families and foster families. Comprehensive services provide all those involved in adoption with support and counseling, before, during and after placement.	www.adoptionresources.org
International Adoption Resources	International Adoption Resources (IAR) is an unbiased resource center for hopeful adoptive parents. IAR offers a wealth of information to provide prospective adoptive parents with the insights they need to make informed and educated international adoption choices. In addition to education, IAR has established both grant and corporate partner travel programs to alleviate the financial barriers to international adoption.	www.theiar.org
Nemours: Kids Health	Comprehensive resource list related to genetics and genetic counseling.	www.kidshealth.org
March of Dimes	March of Dimes researchers, volunteers, educators, outreach workers and advocates work together to give all babies a fighting chance against the threats to their health: prematurity, birth defects, low birthweight.	www.marchofdimes.org

Parent and caregiver resources

Organization	Description	Website
Parents Helping Parents (PHP)	Supports, educates and inspires families and the community to build bright futures for youth and adults with special needs.	www.php.com
Association for Successful Parenting	Is 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.	www.achancetoparent.net/
The National Alliance for Caregiving (NAC)	Is 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.	www.caregiving.org
Parent to Parent USA	Provides emotional and informational support to families of children who have special needs.	www.p2pusa.org
Eldercare Locator	Connects individuals 60+ and their caregivers with the local Agency on Aging Family Caregiver Support Program, which provides respite assistance, support groups and other services for caregivers.	www.eldercare.acl.gov

Respite services

Organization	Description	Website
Epilepsy Foundation	The Epilepsy Foundation offers respite care through its local programs.	www.epilepsy.com/local
ARCH National Respite Network	Assists and promotes the development of quality respite and crisis care programs in the United States; to help families locate respite and crisis care services in their communities; and to serve as a strong voice for respite in all forums.	www.archrespite.org

Mental health resources

Organization	Description	Website
Mental Health America (MHA)	<p>MHA is the nation's leading community-based nonprofit dedicated to addressing the needs of those living with mental illness and to promoting the overall mental health of all.</p> <p><i>Notable Article:</i> <i>Mental Health Resources for Caregivers</i></p>	www.mhanational.org
NAMI	<p>A mental health organization focused on building better lives for Americans affected by mental illness.</p> <p><i>Notable Sections:</i> <i>Your Journey</i> <i>Support & Education</i></p>	www.nami.org
GLMA Health Professionals Advancing LGBTQ Equality	<p>National organization committed to making sure there is health equality for the LGBTQ and all sexual and gender minority (SGM) individuals as well as LGBTQ/SGM health professionals in their environments.</p>	www.glma.org
Mental Health America (MHA)	<p>Dedicated to helping the needs of those living with mental illness and also promote the overall mental health of everyone.</p> <p><i>Notable Section:</i> <i>LGBTQ+ Communities and Mental Health</i></p>	www.mhanational.org
National LGBTQIA+ Health Education Center	<p>Provides educational programs, resources and consultations to health care organizations with the goal of optimizing quality, cost-effective health care for lesbian, gay, bisexual, transgender, queer, intersex, asexual and all sexual and gender minority people.</p>	www.lgbtqihealtheducation.org



8737 Colesville Road, Suite 400 | Silver Spring, MD 20910
800-225-6872 | info@tscalliance.org | tscalliance.org

REPRODUCTIVE DECISION MAKING AND TSC

Deciding to start a family or grow an existing family is a life-changing decision, full of excitement and hope. However, if your family medical history contains a diagnosis of tuberous sclerosis complex (TSC), that excitement and hope can give way to fear and anxiety. Fortunately, geneticists can support families on reproductive decision-making.

The geneticist, along with their team of genetic professionals, tries to meet three goals with the family: diagnosis, prognosis, and recurrence risk. For the purposes of reproductive decisions, when the diagnosis of TSC is already known within a parent or family member, recurrence risk is often the most crucial information a family is seeking. A recurrence risk means the risk (or odds) that a subsequent child will be affected with the same condition.

In the case of diagnosing TSC, the genetic professional's job is to convey the genetic facts about TSC. TSC is a genetic disease caused by a change in the *TSC1* or *TSC2* gene that is causing it to no longer work. The non-working gene can then be passed on to future generations. However, it is important to remember that approximately two-thirds of the time when a child is diagnosed with TSC, neither parent has TSC.

If the change in *TSC1* or *TSC2* that causes TSC in a family member can be detected and identified, further testing can be performed on other family members or used for prenatal testing.

Pregnancy and TSC

Many women with TSC have normal, healthy pregnancies. However, there are some potential health complications to take into consideration when a woman with TSC becomes pregnant, including an exacerbation of her disease particularly related to lung and kidney involvement. Some women with TSC also have a disease called lymphangiomyomatosis (LAM) that involves a bundling of muscle cells within the lung that block air, blood, and lymph vessels, thus affecting the exchange of oxygen within the lungs. For reasons not clear, this condition often worsens during pregnancy causing potentially dangerous complications for the mother and baby. Additionally, there is evidence that angiomyolipomas (a TSC-related kidney tumor) increase in size during

Hope no matter how complex

pregnancy, leading to renal complications. Therefore, it is critical that women know their renal status prior to pregnancy.

Reproductive Options

Reproductive decision making is one of the most sensitive topics a family must discuss, and the choices made are based on a family's beliefs, values, and faith. The TSC Alliance does not advocate for specific options but is committed to sharing the information so families understand all of their possibilities and can make informed choices. It is important to consider that some of these options are still relatively new and very expensive. Often, health insurance will not provide coverage for fertility treatments. If it does, many times the coverage is very limited. Each alternative needs to be considered within a legal and ethical context. Attorneys specializing in family law, perinatologists, reproductive endocrinologists, and geneticists offer professional guidance in supporting families to make the choices right for them.

Prenatal Diagnosis

When the change in the *TSC1* or *TSC2* gene is known, testing can be performed during the pregnancy to evaluate whether the fetus will have TSC. This can be done by one of two techniques: chorionic villus sampling (CVS), which is usually performed between weeks 10-12 of pregnancy; and amniocentesis, which is usually performed between weeks 15-18 of pregnancy. Both carry a small risk of miscarriage. The risk varies depending on the center performing the procedure.

Ultrasounds can also sometimes detect findings of TSC in the fetus; however, they cannot be used to rule out TSC.

Knowing whether the baby has TSC during the pregnancy can be helpful to some families. Some find it helpful so they can prepare for medical care, such as where they will deliver and what doctors they will see. Others also find it helpful emotionally. Although a very difficult decision, and depending on circumstances, some families will choose not to continue the pregnancy.

Prenatal diagnosis can give information about whether the baby has TSC; however, at this time, it cannot be used to predict what symptoms of TSC the baby will have (for example, if he/she will have seizures).

Preimplantation Genetic Diagnosis

When the change in the *TSC1* or *TSC2* gene is known, another option is preimplantation genetic diagnosis (PGD). PGD is technology used to identify a specific genetic change in an embryo created through in vitro fertilization (IVF) before implanting the embryo into the uterus. The purpose of PGD is to identify an unaffected embryo for implantation, thereby reducing or virtually eliminating the risk of having a child with certain genetic diseases.

There are different techniques used for certain indications and varying by center. In general, PGD involves extracting genetic material from the embryo for analysis. Removing

a cell from the embryo for genetic analysis does not hurt the embryo. The embryo compensates for the removed cell and continues to divide normally. This genetic material is then used to determine if the embryo has the change in the gene of interest.

Families with a high risk of having children with a genetic disorder or chromosome abnormalities and who wish to avoid elective pregnancy termination or to prevent the birth of an affected child following prenatal diagnosis may consider PGD. Performing genetic diagnosis prior to implantation of the embryo may reduce the potential for termination of affected fetuses diagnosed by prenatal testing.

There is no difference in pregnancy rates for couples going through IVF and PGD and couples doing IVF alone. The rates are age dependent, but as a general rule it is 30-40% per IVF cycle.

Adoption

Adoption is the process of taking a child into one's family and building a parent-child relationship. This relationship offers all the same rights and privileges of one's biological child. Steps to a successful adoption vary within the United States and internationally. Families typically work through private adoption agencies, social service agencies or religious organizations. Adoptions are legalized through the court system.

Surrogacy

Surrogacy is a method some couples choose as a reproductive option to create a family. It involves a legal contract whereby a woman agrees to become pregnant for the purpose of gestating and giving birth to a child for others to raise. The surrogate mother may be the baby's biological mother (use of the surrogate's eggs) or may be implanted with someone else's fertilized egg.

Egg and/or Sperm Donation

When a couple is concerned about potential risk of passing on a genetic mutation through her eggs or his sperm, options are now available using donor eggs and/or sperm. Donors are available through infertility clinics and private agencies. Egg donation is done in combination with in vitro fertilization (IVF). Sperm donation can be used in combination with IVF and intra-uterine insemination (IUI).

Resources

www.resolve.org: Provides education, advocacy, and support for people facing infertility, multiple miscarriages or having a difficult time with conception

www.sart.org: SART is the primary organization of professionals dedicated to the practice of assisted reproductive technologies (ART) in the United States. The mission of the organization is to set and help maintain the standards for ART in an effort to better serve our members and our patients.

www.adoptionresources.org: The primary purpose of Adoption Resources is to serve the best interests of children, so that each child will be raised in a permanent and loving family. Adoption Resources, strives to provide services that protect the dignity of children, birth parents, adoptive families, and foster families. Comprehensive services provide all those involved in adoption with support and counseling, before, during, and after placement.

www.theiar.org: International Adoption Resources (IAR) is an unbiased resource center for hopeful adoptive parents. IAR offers a wealth of information to provide prospective adoptive parents the insights they need to make informed and educated international adoption choices. In addition to education, IAR has established both grant and corporate partner travel programs to alleviate the financial barriers to international adoption

www.affordingadoption.com: Comprehensive resource list for people seeking financial assistance with adoption costs

www.kidshealth.org: Comprehensive resource list related to genetics and genetic counseling

www.marchofdimes.org: March of Dimes researchers, volunteers, educators, outreach workers and advocates work together to give all babies a fighting chance against the threats to their health: prematurity, birth defects, low birthweight.

<https://ghr.nlm.nih.gov/>: Genetics Home Reference provides overview of genetics, genes, chromosomes, and specific genetic disorders including TSC.

Updated by Hope Northrup, MD and Laura Farach, MD, University of Texas Health Science Center at Houston, August 2021.



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).

Hope no matter how complex



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. 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. Ask for help with laundry. 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.?



Aging Caregiver Checklist

This checklist can help you organize your thoughts as you prepare for a healthy and smooth future while ensuring your loved ones have everything they need to thrive. This list will help you get organized but should not be considered as a substitute for legal and financial planning advice. **Your first step on this checklist is to investigate an agency/consultant to work with for any formal paperwork.**

Recommended steps to life and future planning

1. Determine the needs of the individual with a disability in the following areas, being sure to think of both present and future needs.
 - a. Vocation and education
 - b. Housing
 - c. Medical care
 - d. Recreation, leisure, and hobbies
 - e. Legal needs
 - f. Daily living assistance
 - g. Emotional needs
 - h. Other areas

TIP: Consider, realistically, how much family, friends, and others are willing and/or able to assist an individual now and in the future. Paid life planning professionals can help with these steps. To the maximum extent possible, involve the individual with TSC in developing and implementing each of the life planning steps.
2. Write a "Future Care Plan" (also called a "letter of intent"). This describes your hopes and expectations in writing and provides guidance for future care providers.
3. Recommend future advocates or guardians – people who will visit (as appropriate) and ensure your wishes are carried out.
4. Create a family financial plan that addresses the needs of the individual with TSC, while also providing for the financial objectives of other family members/caregivers.
5. Select a combination of resources that will provide adequate funds for that person's lifetime. This will include government benefits, family assistance, settlements, inheritances, savings, investments, and life insurance.
6. Work with an attorney who specializes in special needs estate planning. Have the proper wills and special needs trust established so resources now, and in the future, protect government benefits and provide supplemental income.
7. Choose a number of successor trustees from relatives and friends or select a corporate trustee to manage the trust funds in the future.
8. Meet with all persons involved to review the plan.
9. Store the life plan, financial plan, and legal documents in a safe place.

10. Review the plan at least once a year. Update the future care plan (from step 2), financial plan, and legal documents as necessary.

Estate and financial planning: quick guide

The topics mentioned in this section are discussed at greater length in the [Adult Guide of the Life Stages Program](#). Develop a solid working relationship with a long-term care provider (i.e., an internist or gerontologist) who can provide insight and support while making these decisions.

- Special needs trust or living trust
- Living will (conservatorship)
- Advanced medical directive
- Healthcare coverage (including Medicaid coverage)
 - Resource: The Arc [Center for Future Planning](#)[®]
- Housing arrangement options, including a transition plan
 - At home with or without assistance
 - Group home with constant (24/7 × 365) supervision
 - Independent living or semi-independent living (e.g., inter-generational communities)
 - Institutional residency

Emergency network: quick list

These are some topics to consider when putting together an emergency network or action plan should anything happen before future plans are firmly in place.

- Contact information for key medical team members:
 - Primary care provider
 - Key specialist(s)
 - Pharmacy
 - Social worker/coordinator if applicable
- Medication list
- “Go bag” that contains emergency supplies for a 24-hour stay at away from home
- Non-medical “who to contact” list
 - Family, friends, or others who may be able to provide support during an emergency (and contact information)

Section Nine: Difficult conversations and end of life issues

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 experience of 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.

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 ensuring families are informed 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 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

1. Control your seizures. Have regular discussions with your healthcare provider if seizures are not controlled.
2. 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.
3. Have a seizure action plan. Make sure to update frequently and give it to others who would need to utilize it.
4. 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.
5. 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.
6. Avoid drugs and alcohol. These can interfere with your seizure medication or lower your seizure threshold.
7. 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.

Advanced decision making

Advance decision making is a process in which you can proactively plan out your or your loved one's wishes on end-of-life treatments and interventions, sometimes called an "advance directive." Ideally, this should be planned or discussed before it becomes medically urgent. Advance directives provide a roadmap of how to carry out difficult tasks during a difficult situation. These directives can provide peace and comfort to those who have to act in the best interest of their loved ones. An advance directive is a legal document that should be part of your estate planning. The difference between legal documents and living wishes can both be honored in a time of need.

Having an advance directive in place is especially important when caring for someone with special needs. Thinking ahead proactively about these difficult choices can help avoid stress when emotions and doubt threaten to drive decision making. It's normal to experience many emotions including fear, doubt and guilt during this process. It is important for the caregivers to have a clear understanding of their loved one's baseline behaviors and quality of life. If your loved one has physical and/or cognitive disabilities, it is incredibly helpful to your healthcare team if you create a plan that outlines the best ways to communicate with your loved one should he/she be admitted to a hospital.

It's important to understand you are not alone during this time. It feels wrong to accept loss, but there's also a great act of kindness in helping your loved one pass with dignity. Deciding to have these difficult conversations earlier might be hard, but it is much harder to think clearly during a crisis. Having a plan in case of a serious illness is okay and will look different for each family. If your loved one is younger, it may be better to discuss only one or two topics at a time.

As you process and navigate this phase remember all decisions including hospice and do-not-resuscitate (DNR) orders can be revoked at any time. Having a plan in place doesn't mean you're irreversibly committed to each decision – it is essentially a starting place meant to provide support, not stress, and it can be adjusted appropriately according to your situation.

Advanced decision making planning considerations

Take the time to develop a care plan for current and possible future challenges. The purpose of planning is to help guide your decisions in the event of an acute situation in a stressful time when emotions, fears, doubts and guilt can take over and cloud your judgement. NO decision is permanent, so if you choose to change your decision at any given time along any healthcare journey all prior decisions can be revoked if you are the legal caregiver or guardian. Those who have legal medical power of attorney or conservatorship/guardianship would be the legal representative to make end-of-life decisions and carry out wishes that have already been discussed, which can offer comfort to those responsible during these times.

This plan helps identify the decision making process for the entire caregiver team. It is important to understand the development of the plan is a process, because not all situations are relevant and can be addressed over time. The plan is unique to the caregiver's personality and the loved one's condition. There are no right or wrong decisions. It is never too early to discuss these difficult situations.

Things to consider to be a part of a legal document or discussed with the caregiver team (family) include:

1. Nutrition, food and hydration
 - Situations where alternative routes include a nasogastric tube or more permanent PEG-tube. These are both invasive, yet minor procedures. Both are performed at the bedside. Once you or your loved one has recovered, these can both be removed and typical nutrition feeding routes can be resumed.
 - Situations where decisions of withholding certain nutrition interventions. This is more common during irreversible situations when the end of life is being discussed. During the dying transition, the desire or metabolic needs for food or water change. It is important not to force food or water during this time. Allow the person to decide the timing and amount of food consumed. It is about comfort not nutrition. Forcing food can have unfortunate consequences such as aspiration (food accidentally goes down airway).

2. Symptom control and management

- Physical areas:
 - o Pain regimen: How to identify pain – is it verbalized or through facial or body cues? What is acceptable and safe? When is comfort your main priority over treatment? Under some situations pain medication will be withheld to maintain basic life function. There is never a “giving up” period. Care goals change based on all the information in specific, unique situations.
 - o Infection treatment and management: Are there situations during care where you would not want antibiotics, wound therapy, surgical debridement, wound vacs etc.?
 - o Acute issues such as breathing and neurological function. Are there situations where you would not want advanced life support?
 - o Blood Transfusions: Is this acceptable treatment or are there religious or personal beliefs that restrict this option?
- Psychological areas: How to provide comfort and support especially if your loved one is nonverbal during difficult times? What is a source of comfort for your loved one, such as a special blanket, pillow, stuffed animal or book?
- Spiritual support areas: Are there spiritual needs necessary during difficult times?

3. Resuscitation/“life support” Interventions

- Do not resuscitate/do not intubate (DNR / DNI): This means there will be no medical interventions by medical personnel if core basic functions have stopped such as no longer having a heartbeat or absences of breathing.
- Full code: This is where all life-saving interventions would be completed by medical personnel in the event the heart or breathing stopped. These interventions include chest compressions (pumping on the chest wall), intubation (a breathing tube), defibrillation (shocking with electric pads) and medications (given in efforts to reverse fatal arrhythmias, causes or in hope to regain a heartbeat).
- These interventions can be unique and specific to you or your loved one. These interventions can be all the above, none or certain in-between varieties based upon your wishes. You have the right to use a full code (all elements above carried out), or a partial code where you can select which elements you do and do not want carried out. You can also just request to be a chemical code, where life-saving medications would be pushed through an IV to attempt to restart the heart in the event the heart stops. You can also request chest compressions only.
- It is important to understand these decisions can be reversed at any time. Having a plan in place or having discussions with another caregiver regarding these difficult conversations can change throughout your journey. Adapt accordingly. Empowered decisions are best made with those who are knowledgeable about all the options so you can make the best choices for your loved one.

4. Acute dialysis: This intervention can be done due to an acute illness in someone who is susceptible to kidney failure. However, this can also occur in situations where the body has started to shut down and the kidneys are no longer processing in a way to sustain life. It would be important to understand the situation to be able to navigate if this intervention should occur. Regardless, there are some who might feel this option would not be in the best interest of their loved one regardless of the etiology. All decisions are unique and should never be compared to others even with the same rare disease or other diagnosis.

5. Other medical interventions and procedures: It is impossible to cover all options, but the main objectives will always remain the same. The type of questions you would want to ask yourself include:

- What is the purpose of this intervention or procedure?
- Will this fix the current issue or relieve stress?
- What is the benefit of this procedure?
- Is this a temporary fix or will this procedure or intervention need to be repeated and if so, what does that look like.
- What is the risk of these procedures?
- Will this intervention be long term?
- Does this intervention change our quality of life?
- What are the added barriers if we carrier out this intervention?
- Is this a sustainable option? How long could this be feasible for the family dynamic?

Difficult situations can provoke emotions due to different points of view in families. Be open and compassionate to each other during these discussions and keep the overall goal about the loved one you all care about centered.

Transition phases in caregiver roles for an adult child with TSC, such as an aging parent to an adult sibling or spouse, should consider having this discussion with the adult sibling/child as an equal part of the process. If these conversations need to be addressed but you feel it will cause issues within a family dynamic, consider reaching out to a local counselor, clergy or palliative care service to help moderate these difficult talks. Remember, you are not alone during these times.

Acute decision making planning considerations

The following considerations about acute care decision making will help you navigate the very difficult questions that arise during these times. These considerations will help you understand your options with a very generalized approach. We hope going through this example will empower you and provide direction when planning your loved one's care.

Initiate a meeting to establish (and update) a care plan

Request a meeting to discuss family goals for care – include the attending physician, nurse practitioner, nurse manager, social worker, case manager, other key healthcare team members and other specialists. This meeting helps develop an agreed plan of care.

- This type of meeting can be held as often as possible and is typically arranged by the unit or floor social worker or case manager.
- You can request to have outside services or advocates such as a family member, patient advocacy representative or outpatient provider. These participants will help you better understand the discussion and articulate your thoughts to the medical team. Often these participants are on the phone.
- This is a forum to discuss any issues in care, expectations of care, or interventions that could or should be carried out.

Discussion topics to consider with your healthcare team

- What is the acute illness (reason for admission) and is it reversible?
 - How long will this acute illness last with the current care plan?
 - What to expect in “good” and “bad” days
 - What does it mean if there are not any changes in symptoms? Would that be expected, or is that a sign of something else that might require additional intervention?
- What are the underlying comorbidities that might increase the chance of a poor outcome?
 - Are those underlying issues worsening during this acute illness?
 - Neuro-relevant questions:
- Has there been constant status epilepticus? If so, how long?
- What are the underlying cognitive changes that are being seen by the medical team either by diagnostics or by physical exam?
- Are there changes in behavior or communication?
- Kidney questions:
 - Are renal issues acute or are the kidneys functioning stably? NOTE: If your loved one is receiving organ-saving acute hemodialysis, it is not considered to be end-stage renal disease unless the kidneys have not started to work after six months.
 - Is this a quick fix that will most likely resolve in an outpatient setting or are there other severe acute issues such as other organ failure?
- Lung questions:
 - Are there respiratory changes?
 - Is oxygenation required to sustain acceptable levels?
 - Does their breathing look labored, difficult, uncomfortable?
- Other health-related issues such as heart attacks or stroke are all important to consider.
 - How does this change the baseline and function of your loved one?
 - Does it impact the quality of life that you seek for them?
- Will any of the comorbidities permanently affect the quality of life?
 - This is always difficult in critical care as “time” is usually the only indicator if one will improve or not.
 - Remember: At baseline, your loved one has an incurable disease, and some healthcare providers might not understand goals of care and what is an acceptable change from this baseline. NOTE: It is very important to have a clear established understanding with your healthcare team of what is the ongoing quality of life that is effective and OK for your loved one.

- Are pain and quality of life being balanced?
 - o Those who have loved ones who are nonverbal or with a lower cognitive function can find these times more difficult during an acute situation. Having a discussion with your healthcare team or having a list available to discuss ways to interact, communicate, and pick up on their verbal and non-verbal cues as to whether they are hurting is very important to the bedside nurses as well as the overall healthcare team.
 - o Resources such as communication boards or a quick reference guide (e.g., a poster board to hang on the wall that shows certain types of verbal sounds or movements your loved one makes when in distress or pain) is invaluable to the medical team and should be discussed as soon as admitted.
- Understanding the risk of death if changes are not implemented.
 - o EXAMPLE: Is a breathing tube or other form of life support required to maintain life? This can be a very difficult question, and the younger the age the more difficult it is to talk openly with parents from a healthcare perspective.
 - o If you are faced with end-of-life consideration, medical professionals (e.g., hospice team, pain management team) are available to assist during these times. These teams typically include a physician, nurse practitioner, nurse and a social worker.
 - o Remember not all palliative care is hospice, but all hospice is palliative.
 - o If you are faced in a situation involving difficult scenarios and difficult life choices, always consult the attending medical doctor or team – you are not alone in this journey.
- The COVID-19 pandemic has created many challenges in medical facilities.
 - o Due to the complex changes, many institutions have been inconsistent and unpredictable with visitation policies.
 - o The most common question is “how will I know if my loved one’s needs are being met with compassion and quality” imposed by COVID-19 restrictions?
- Ask up front about visitation policies. Depending on the state, Title III of the ADA does require hospitals to provide accommodations.
- Most facilities do make accommodations to allow one support person to always remain at the bedside.
- This does change depending on the higher level of care needed such as an intensive care unit, and this can change and influence the accommodation that you might’ve had in a different setting of the same hospital.
- If you are found in this situation it is best to speak with the intensive care nurse manager, palliative care nurse, social worker or case manager to discuss a family intervention plan.

Despite clear and direct communication, you might still find yourself in a situation where you do not feel like your voice is being heard or the needs of your loved one are being met. If that is the situation, the following steps may help expedite your concerns and increase resolution.

- Communicate directly with the bedside nurse.
- Discuss care expectations with the nurse manager.
- Repeat these steps as necessary during a hospitalization. If you still do not feel you are being heard or interventions are not being carried out at any point in this process, you can contact the hospital patient advocate or liaison. This can occur at any stage of the hospitalization including in the emergency department. If things continue to not go well and you need to expedite care, you can always reach out to the chief medical officer or, depending on the issue, request an ethics committee consultation. It is best to try to resolve any conflict prior to those steps, but all situations are unique and understanding your options will help empower you to have a sense of direction during the most difficult circumstances.

If you decide to have a family meeting, it’s always best to have a written agenda with your concerns and questions you want addressed. Don’t be afraid to speak up. Don’t be afraid to have an advocate who is slightly removed emotionally from the situation to make sure your objectives are being carried out, so you also have a clear picture of what is going on given the current barriers and restrictions.

If you are alone but would like to express your wishes in writing, ask the unit security or bedside nurse to make a copy of your agenda so each person attending the meeting can read it themselves, thus putting less pressure on you. Another key person to consider having present is a chaplain, clergy or religious leader (the hospital can help you connect if you don’t have one in mind) for moral support for you and who can moderate discussion if it becomes intense between you and the healthcare team.

Hospice care

What is hospice?

Hospice care is medical care for individuals who have experienced a sudden decline in their health or have received a terminal diagnosis. The life expectancy is six months or less and the attention is to focus on aggressive care management to optimize comfort and quality of life for the time that is left. The hospice care team includes medical professionals trained to address the needs of the individual at this stage. This can mean anything from pain management as well as physical, psychosocial and spiritual needs as they become the focus of care with less emphasis on the disease.

For those who have outlined an advanced decision making plan, the hospice process is usually a by-product of your loved one not responding to current medical interventions that may have worsened overall health and well-being such as sepsis, progressive pneumonia, resistant urinary tract infections and, for some, status epilepticus that has left your loved one in a coma.

Understanding what options and interventions you want or what you want your loved one to receive before an acute episode does help minimize some of the emotions; however, this is still a very emotional and difficult decision to make regardless of the loved one's age. For those who have outlined interventions that are not wanted, such as hemodialysis, hospice services are an excellent option to offer high quality of life for them to continue to enjoy the remainder of their lives where they want, how they want and with who they want.

How does hospice care work?

Hospice can be provided in multiple types of settings – in the home or at a facility, such as a hospital, nursing home or hospice center. Acute hospice options are also available, which are usually within the acute care setting (inpatient) when care is being withdrawn or deescalated to allow time and interventions to be set up at one of the previous options. Depending on your needs, some of these options might not be offered. If you find yourself in this situation, most hospitals have hospice coordinators who you can discuss your unique needs with to align to which option is best for your loved one.

How do I find hospice care?

Once it is determined you or your loved one qualifies for hospice, hospice care can be initiated within the inpatient or outpatient setting. Depending on your overall situation, goals of care, resources and needs, your local provider or your primary hospital provider (the doctor you are admitted under) will help initiate this process. It is important to know that the Centers for Medicare & Medicaid Services (CMS) require any option/ agency that provides care in your area is offered to you as the family. Now, most families defer to an agency suggested by their healthcare provider, but if you want to know all options then you have that as a right. The final decision is yours. It is strongly recommended, if time appropriate, to visit any hospice facility or long-term care facility that offers hospice agencies prior to being discharged.

Who pays for hospice care?

Depending on the type of insurance, you or your loved one will determine the cost of hospice. Medicaid and or Medicare typically pays for 100% of hospice services, which include the medical team, therapy, medications and supplies needed. Commercial insurance still offers hospice benefits but can be slightly different than those who qualify for CMS support that are covered by Medicare Part A. Depending on the location of hospice, such as a hospice house or skilled nursing facility, other charges can accrue. Some insurances will cover the full cost, while others have a daily fee. If you are exploring these options in a non-acute situation your case manager, health benefits managers or appointed personnel through human resources would be the best resources to understanding your hospice benefits. If you are in an acute situation, the best option would be to discuss these questions with the hospice coordinator at the agency of choice, the hospital's hospice coordinator or your social worker/case manager.

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.

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 2,000 people with TSC are enrolled in the Natural History database, and more than 800 people have contributed biosamples. The biosamples and data about individuals who provided samples 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. In coordination with a national mobile phlebotomy service (Phlebotek), the TSC Alliance can collect blood samples from anyone in the United States with TSC. 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. 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.

Tissue samples

If an individual with TSC is having surgery to remove any part of 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. The TSC Alliance is also able to collect cord blood and placental tissue. For postmortem 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. **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.

Section Nine Appendix

Grief and bereavement resources

Organization	Description	Website
The Dougy Center	Provides grief support in a safe place where children, teens, young adults and their families can share their experiences before and after a death. It provides support and training locally, nationally and internationally to individuals and organizations seeking to assist children who are grieving.	www.dougy.org/grief-support-resources
Hope for the Bereaved	Dedicated to helping children, teens and adults by providing support groups, a monthly newsletter and counseling at no charge.	www.hopeforbereaved.com
The University of Maryland Brain and Tissue Bank (UMBTB)	The UMBTB collects, stores, distributes donated brain and other tissues for research to help improve the understanding, care and treatment of individuals with developmental, neurological and movement disorders. Direct Email: btbumab@som.umaryland.edu Call for imminent need: 1-800-847-1539	www.medschool.umaryland.edu/btbank/

Hospice services

Organization	Description	Website
Hospice Foundation of America	Educates the public and health care professionals about death, dying and grief.	www.hospicefoundation.org
National Hospice and Palliative Care Organization (NHPCO)	Works to expand access to a proven person-centered model for healthcare-one that provides patients and their loved ones with comfort, peace and dignity during life's most intimate and vulnerable experiences.	www.nhpco.org
National Association for Home Care & Hospice	Promote, protect, and advance the highest quality health care at home.	www.nahc.org



Hope no matter how complex™

8737 Colesville Road, Suite 400
Silver Spring, MD 20910-4487
(800) 225-6872 | tscalliance.org | [@tscalliance](https://www.instagram.com/tscalliance)