



# Navigating the early years of TSC

An information guide to help parents and caregivers navigate tuberous sclerosis complex (TSC) from in utero diagnosis to age five.



## 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](http://www.tscalliance.org/tscnavigator)) and focus on providing you with proactive material to help navigate the medical and non-medical aspects of this journey specific to the defined age group. While each developmental phase brings its own share of excitement and challenges, our guides are to complement your TSC journey and to empower you to live your fullest life.

### Navigation guides across the lifespan

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

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

# Contents

<b>Section One: My child has TSC, now what do I do?</b> .....	<b>3</b>
What is tuberous sclerosis complex? .....	3
Where do I start? .....	3
Where do I go? .....	3
How do I get connected? .....	4
Section One Appendix.....	5
<b>Section Two: Navigating the medical journey</b> .....	<b>7</b>
Understanding the diagnosis .....	7
Clinical manifestations .....	7
TSC diagnostic criteria.....	7
2021 TSC diagnostic criteria updates .....	8
TSC genetics.....	8
Clinical management and your healthcare team .....	8
Staying organized.....	9
Medical challenges.....	9
Pharmacy coverage .....	9
Prior authorization issues.....	10
Insurance barriers .....	10
Tips to mitigate issues.....	11
Medication assistance .....	11
Clinical trials and research opportunities.....	12
What is a clinical trial? .....	12
Why participate in a clinical trial? .....	12
Who can participate in a clinical trial?.....	12
What are the benefits and risks of participating in a clinical trial?.....	12
Where do I find a clinical trial for TSC? .....	12
Where can I get more information about clinical trials? .....	12
Health insurance and medical benefits .....	13
Section Two Appendix .....	13
<b>Section Three: TSC-associated neuropsychiatric disorders (TAND)</b> .....	<b>30</b>
What is TAND? .....	30
Assessment of cognitive and behavioral health issues .....	30
Therapies and services.....	30
Documenting behaviors .....	31
Section Three Appendix.....	32
<b>Section Four: Legal rights under disability law</b> .....	<b>34</b>
Americans with Disability Act (ADA) .....	34
Individuals with Disabilities Education Act (IDEA) .....	34
Section 504 .....	34
Legal assistance .....	35
Family and Medical Leave Act (FMLA) .....	35
Air Carrier Access Act .....	35
Telecommunications Act.....	35
Section Four Appendix .....	36

<b>Section Five: Early intervention</b> .....	<b>37</b>
What are Early Intervention services under IDEA? .....	37
Who do I first contact for help? .....	37
What is an Individualized Family Service Plan? .....	37
Early childhood transition .....	37
Section Five Appendix.....	38
<b>Section Six: Educational rights</b> .....	<b>39</b>
Services under IDEA.....	39
School evaluations.....	39
Developing an Individualized Education Program (IEP) .....	40
Preparing for an IEP meeting.....	40
Understanding IEP components.....	41
IEP requirements.....	41
Present Levels of Academic Achievement and Functional Performance (PLAAFP) .....	42
Measurable annual goals .....	42
Additional services in the school setting .....	42
Section Six Appendix .....	43
<b>Section Seven: Assistive technology (tools and devices)</b> .....	<b>45</b>
What is assistive technology? .....	45
What is the purpose/benefit of assistive technology?.....	45
How do you fund these services?.....	45
Section Seven Appendix .....	46
<b>Section Eight: Self care and family dynamics</b> .....	<b>47</b>
Caregiver mental health.....	47
Signs and symptoms of caregiver burnout .....	47
Continuum of grief.....	48
Family discussions.....	49
How to talk to your child about TSC.....	49
How to talk to your unaffected child(ren) about TSC .....	49
Respite care .....	50
Section Eight Appendix.....	50
General family resources.....	50
Parent and caregiver resources.....	51
Sibling resources.....	51
Mental health resources.....	52
Respite services .....	52

## Section One: My child has TSC, now what do I do?

### What is tuberous sclerosis complex?

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

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

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

### Where do I start?

Receiving a diagnosis of a rare genetic disease for your child is overwhelming. Depending on the findings, it is important to have ongoing conversations with your healthcare team about what to expect. Often, a TSC diagnosis can be sudden, which can be even more overwhelming to process. For example, there can be an immense amount of testing and specialist visits to coordinate all at once, which might feel like trying to drink from a firehose.

Other times, TSC might be suspected, which can lead to further evaluation after a non-threatening feature like a skin spot (such as an “ash leaf” or a Shagreen patch) is identified. Regardless of the presenting feature, once a diagnosis has been made or suspected it is important for your child to undergo a thorough evaluation. Sometimes it might be appropriate to hold off on a thorough TSC-specific evaluation until any acute symptoms are stabilized. For example, if your child is experiencing infantile spasms, it might make sense to get them under control before doing an extensive dermatological exam.

Key factors in evaluating TSC are outlined in the 2021 Updated Diagnostic Criteria (see Section Two). It is important to establish a 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.

### Where do I go?

It is common for medical providers to be unfamiliar with TSC, including neurologists and pediatricians. It is crucial to have a local neurologist and pediatrician involved in your child’s healthcare team. Finding a good fit is essential as these care team members will be your child’s experts, and along with you, be your biggest advocate as you start your TSC journey. During this age span, a pediatric neurologist who is familiar with TSC should be part of your child’s healthcare team. Epilepsy (seizures) is frequently the main manifestation of TSC that leads to the diagnosis but not always. Even if your child presented with a TSC diagnosis and has not shown any signs of seizures, it is still clinically recommended to be evaluated during this phase as signs and symptoms of TSC change rapidly during these years due to normal growth and development. As a parent you want nothing less than the best and connecting with a local or out-of-state TSC Clinic to help guide clinical treatment options is appropriate. If you do not have access to a local TSC Clinic, you can also make an appointment with a pediatric neurologist experienced in epilepsy. Many TSC experts can offer provider-to-provider recommendations and the TSC Alliance’s TSC Support Navigators can assist with this process.

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

## How do I get connected?

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

This journey can feel lonely at times so connecting with another family that has navigated these steps before you can make all the difference. To gain more insight on caregiver selfcare please go to caregiver mental health under the resource section. In addition, plenty of ways exist to help you connect with the TSC community or feel free to reach out to our Support Navigators for assistance.

Finding your local Community Alliance is an excellent place to start. Please visit the TSC Navigator at [www.tscalliance.org/tscnavigator](http://www.tscalliance.org/tscnavigator) for more about a Community Alliance near you.

For up-to-date information about TSC Alliance initiatives, videos, town halls, podcasts, information sessions, research, events and community stories, you can find the TSC Alliance on social media platforms using @tscalliance. If you feel like volunteering, plenty of opportunities exist to align with any specific interest you feel is best suited for you.

### **TSC Alliance social media channels:** @tscalliance

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

## Facebook options

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

Facebook also has other private support pages (not operated by the TSC Alliance) for TSC Mommies and TSC Daddies. To join these private groups, you can search for TSC Mommies ONLY – Tuberos Sclerosis Complex or TSC Daddies and request to join.

## Volunteer opportunities

Volunteering offers an opportunity to connect and engage with other families in the TSC community. There are various ways to participate as a volunteer with a position to fit anyone who is interested. For more information about a volunteer position please go to [www.tscalliance.org](http://www.tscalliance.org) for more information.

## Section One Appendix

### TSC Alliance resources

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

Resource	Description	Website
<p><b>TSC Navigator</b></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><a href="http://www.tscalliance.org/tscnavigator">www.tscalliance.org/tscnavigator</a></p>
<p><b>TSC Now</b></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><a href="http://www.tscalliance.org/tscnow">www.tscalliance.org/tscnow</a></p>
<p><b>TSC Matters</b></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><a href="http://www.tscalliance.org/tscmatters">www.tscalliance.org/tscmatters</a></p>
<p><b>TSC Academy</b></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><a href="http://www.tscacademy.org">www.tscacademy.org</a></p>
<p><b>Webinars and Videos</b></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><a href="http://www.tscalliance.org/webinarsandvideos">www.tscalliance.org/webinarsandvideos</a></p>

## Other notable resources

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

## Section Two: Navigating the medical journey

### Understanding the diagnosis

TSC is a genetic disorder that causes tumors to form in many different organs, primarily in the brain, eyes, heart, kidney, skin, and lungs. Most people with TSC will live a normal life span. Although there is no cure, there is hope. Research has proven that early diagnosis and intervention(s) are key for optimizing long-term outcomes. Advancements in research continue to deliver new and improved therapeutic options. As previously mentioned, 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 TSC community.

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

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

**Possible TSC:** Either 1 major feature or >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

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

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

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

Please reference the specialists you may encounter in this journey in the appendix of this section for more insight. Since TSC is a rare disease, sometimes you will have to travel out of state to see a TSC expert for clinical care or for those interested in clinical trials. The Bcureful Travel Fund 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 TSC Clinics. Please see the TSC 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 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, many laboratory and diagnostic tests 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 is critical with this diagnosis. Building a solid foundation now will make a huge difference during your journey. Learn more on Understanding Medication and Testing, which can be found in the appendix of this section.

Trying to stay organized when you are a parent or a caregiver to an individual requiring various appointments can be overwhelming. It is essential to be organized. Creating a medical binder, or notebook, so 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 a downloadable version go to the TSC Navigator's resources section.

## Medical challenges

### Pharmacy coverage

Prescription drug plans are part of all commercial and government insurance plans in the United States; however, each plan is different per corporation/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 what 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 – 3 usually require some co-pay, and it is not uncommon to have your co-pay cost listed on your pharmacy drug card. Most of these medications are generic, and brand name medications are often higher up on the tier list. If you have Medicaid or Medicare, it is not uncommon for your insurance to cover brand names. Some states have restrictions on how many brand-named drugs you can use per month (this included 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, because each year in January, your progress toward "meeting your deductible" (i.e., the amount of money you spend each year) resets.

Many people utilize copay assistance cards so medications can be affordable; however, under the current copay 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, who 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 what 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 have to initiate the new PA before you run out of medication. Medications requiring a PA will also be listed on your pharmacy drug list (PDL). For those who need an example letter, please check out the Prior Authorization Issues in the TSC Navigator for the most current templates.

## Insurance barriers

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

- The individual being seen MUST have an established primary care provider (who is licensed in the state the individual lives in) who sees them routinely. This primary care provider needs to have seen the individual at least once within the current calendar year BEFORE an out-of-state referral is made. Medicaid coverage for out-of-state referrals is often delayed because:
  - o Your primary care office staff typically need 30 days to request records and be able to review.
  - o Your primary care office staff must justify to your home state's Medicaid why services need to be rendered OUTSIDE 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. Most require annual submission of forms, so insurance does not lapse. Not knowing and having them "termed" or "inactive" will always result in DELAYED care.
- When referred to out-of-state care with Medicaid, ensure ACCURATE information is provided and the out-of-state provider is enrolled with that state's Medicaid program.
  - o Contact the out-of-state clinic prior to the referral being placed to see the provider(s) are enrolled.
    - IF NOT, the out-of-state team will need to complete this enrollment 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 child.
    - Why does this matter? If these individuals are not properly enrolled, claims will be denied, and the institution will not get paid for services rendered.
    - For those with commercial insurance, this may also be an issue; if this is not properly done on the front-end, insurance can deny service 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 7-14 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 (Sabril®), but your plan will only cover generic (vigabatrin). If this substitution is allowed, then call your prescribing providers office to let them know as soon as possible. Never assume your providing office will be notified of this denial in a timely manner.
4. Are compounded medications covered under the current plan for “off-label” use? Compounded medications are ones that undergo a transformation within the pharmacy to change their delivery method – for example, a medication 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 who are still in need of financial assistance. While these programs often limit the total amount that an individual can utilize per year, it is helpful to know what options are available. Co-pay assistance programs are typically for medications that are brand name with no generic equivalent available. Often, your prescribing provider or specialty pharmacist can help direct how to get set up for this type of program, but they cannot do everything for you because personal financial information such as tax returns may be required.

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

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

## 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](http://www.tscalliance.org/clinicaltrials).

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

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

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

## 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.

## Section Two Appendix

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



# 2021 TUBEROUS SCLEROSIS COMPLEX

## DIAGNOSTIC CRITERIA, SURVEILLANCE AND MANAGEMENT RECOMMENDATIONS

### Diagnostic criteria

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

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

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

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

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

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

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

Organ System or Specialty Area	Recommendations	
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. Adrenocorticotrophic hormone (ACTH), synthetic ACTH or prednisolone can be used if treatment with full-dose vigabatrin for 2 weeks has not correlated with clinical and EEG improvement.</p> <p>Antiseizure medications (ASM) for other seizure types in TSC should generally follow that of other epilepsies. Everolimus and a specific cannabidiol formulation are approved by regulatory authorities for treatment of seizures associated with TSC. No comparative effectiveness data exist to recommend ASM, everolimus, cannabidiol, or dietary therapies over one another in specific subsets of patients.</p> <p>Epilepsy surgery should be considered for medically refractory TSC patients at epilepsy surgery centers with expertise in TSC. Special consideration should be given to children at younger ages experiencing neurological regression and evaluation for surgery should be performed at epilepsy surgery centers with experience and expertise in TSC.</p> 
TAND	<p>Perform annual screening for TAND, using validated screening tools such as the TAND Checklist (<a href="http://tandconsortium.org/checklists/">tandconsortium.org/checklists/</a>). Screening may be done more frequently depending on clinical needs. When any concerns are identified on screening, proceed to further evaluations by appropriate professionals to diagnose and treat the relevant TAND manifestation(s).</p> <p>Perform comprehensive formal evaluation for TAND across all levels of TAND at key developmental time points: infancy (0–3 years), preschool (3–6 years), pre–middle school (6–9 years), adolescence (12–16 years), early adulthood (18–25 years), and as needed thereafter.</p> <p>Refer to appropriate professionals for the management/intervention of relevant TAND manifestations. Interventions should be personalized to the TAND profile of each individual and be based on evidence-based practice guidelines/practice parameters for individual manifestations (e.g. autism spectrum disorder, attention deficit hyperactivity disorder, anxiety disorder).</p> <p>Aim for early identification of TAND manifestations and early intervention.</p> <p>Many people with TSC have academic/scholastic difficulties. Therefore, always consider the need for an individual educational program (IEP/IEDP).</p> <p>Sudden and unexpected change in behavior should prompt physical evaluation to look at potential medical causes (e.g., SEGA, seizures, renal disease, medications).</p> <p>Provide psychological and social support to families and caregivers and ensure strategies are in place to support caregiver wellbeing. Continue to provide parent/caregiver education and training about TAND to ensure families know what to look out for in emerging TAND manifestations across the lifespan.</p> 

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



8737 Colesville Road, Suite 400 | Silver Spring, MD 20910  
800-225-6872 | [info@tscalliance.org](mailto:info@tscalliance.org) | [tscalliance.org](http://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?

**Hope no matter how complex**

## Insurance

- Will my insurance cover this test or treatment?

## Lifestyle changes

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

# Master Appointment Schedule

Year: 2022



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

# Doctor Visit Log



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

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

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

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

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

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

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

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

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

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

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

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



## Communication Log

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

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

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

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

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

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

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

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









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

## TUBEROUS SCLEROSIS COMPLEX TRAVEL & LODGING ASSISTANCE PROGRAMS

TRAVEL &  
LODGING  
ASSISTANCE

### What is the purpose of these programs?

These Programs provide patients diagnosed with tuberous sclerosis complex with financial assistance for travel and lodging related to participation in a clinical trial and /or furtherance of treatment at a TSC Alliance-recognized Center of Excellence or TSC clinic.



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



### About the Programs:

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

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

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

## What kinds of assistance are available?

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

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

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

## How do I get more information and apply?

Contact NORD T&L Assistance Program

Monday-Friday 8:30 am – 6:00 pm ET

☎ 203.616.4320

📠 203.349.3199

✉ TSCTravel@rarediseases.org

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

## NORD is Here for You

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

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

©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](http://rarediseases.org). NRD-2131



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

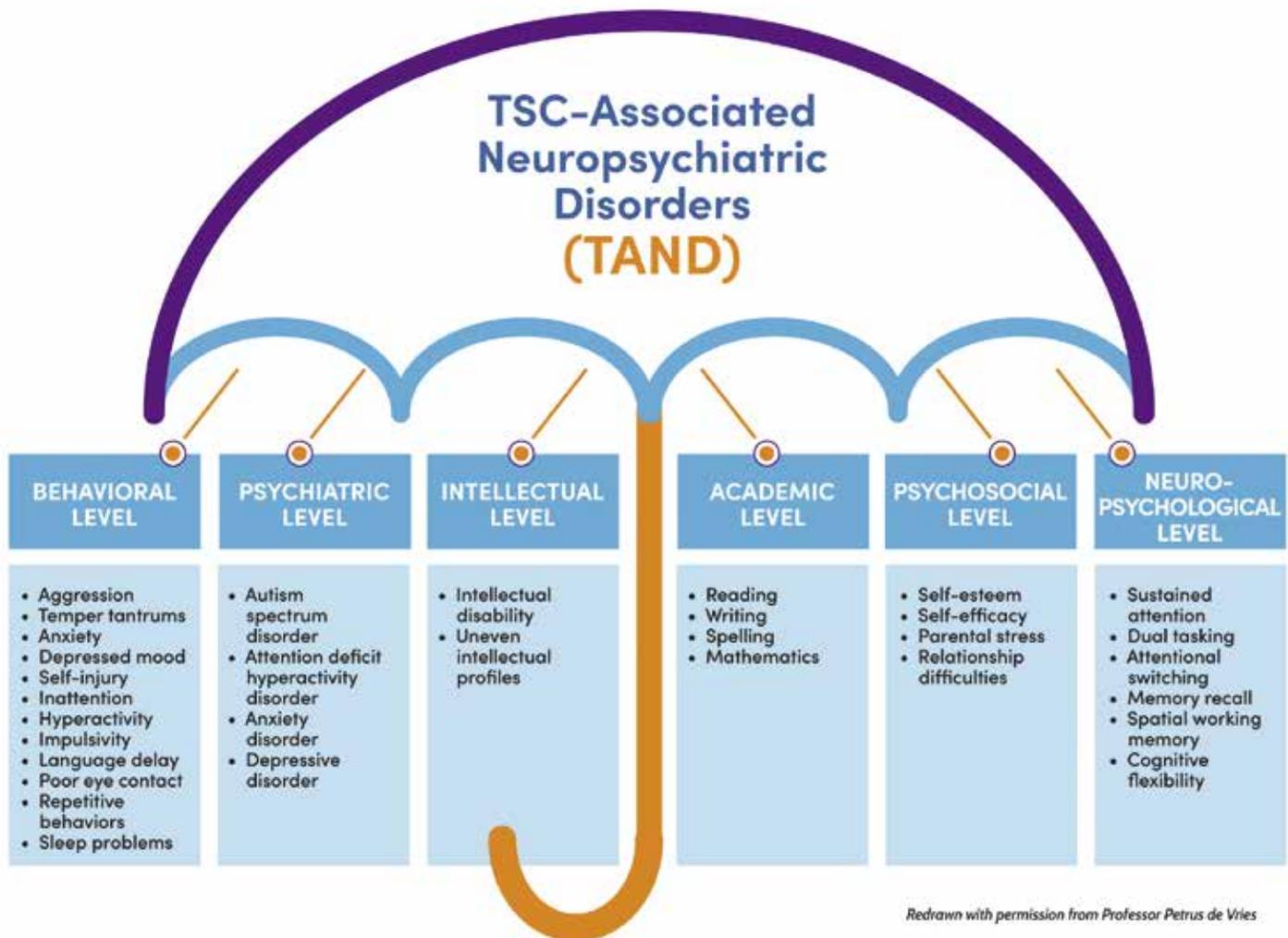
## Section Three: TSC-associated neuropsychiatric disorders (TAND)

### What is TAND?

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

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

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



## Assessment of cognitive and behavioral health issues

In addition to the various physical manifestations of TSC, about 90% of TSC individuals are affected by a range of behavioral, psychiatric, intellectual and neuropsychological complexities. This can include Autism, ADHD, intellectual disability, anxiety and depression. 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.

## Documenting behaviors

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

### EXAMPLE:

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

**Context of the incident:** During dinner Tom was acting up. His sister started laughing at him and he started screaming and throwing his food.

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

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

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

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

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

## Section Three Appendix

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



## Challenging Behaviors Log

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

Explain Incident: Logan was hitting me and throwing various items across the room.

---

---

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

---

---

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

---

---

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

Consequence: Logan lost iPad privileges for the evening and had to help pick up all of the items he threw

---

---

Notes: Will send a note in MyChart to Dr. Krueger about escalating behaviors during transitions

---

---

---

---

## 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 more 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.

### Individuals with Disabilities Education Act (IDEA)

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

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

For more information on the IDEA 2004 visit the U.S Department of Education website listed in this section’s appendix.

### Section 504

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

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

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

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

## **Legal assistance**

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

## Section Five: Early intervention

### What are Early Intervention services under IDEA?

As discussed in Section Four, the Individuals with Disabilities Education Act 2004 (IDEA 2004) is an education law designed to ensure that children with disabilities in the general education setting to the “maximum extent that is appropriate and make meaningful progress through a free appropriate public education (FAPE).”

### Who do I contact first for help?

It is recommended that families who have received a diagnosis of TSC be evaluated for early intervention services as soon as possible. Each state determines which of its agencies will be the lead agency in providing early intervention services for infants and toddlers eligible for Early Intervention services. Visit the Centers for Disease Control and Prevention website for state-specific contact information; you can find its information in the appendix of this section.

When contacting the appropriate agency for your state you will be asked to provide information on the reason for the Early Intervention referral. The case will then be assigned to a service coordinator or case manager that will schedule an intake meeting. Following the intake meeting an evaluation and assessment will be scheduled.

### What is an Individualized Family Service Plan?

After the evaluation, you will meet with the team of professionals to review the results at an eligibility meeting. The guiding principle of the Individualized Family Service Plan – often referred to as the IFSP – is that the family is the child’s greatest resource, and his/her needs are closely tied to those of the family. The best way to support children and meet their needs is to support and build upon the individual strengths of their family. The IFSP is a whole family plan with the parents as the most important part of the IFSP team. Depending on the child’s needs, Early Intervention services might include family training, counseling and home visits; occupational, physical, or speech therapy; hearing loss services; health, nutrition, social work, and assistance with service coordination; assistive technology devices and services; and transportation.

The IFSP will describe the following:

- Present levels of development
- Family information (with parents’ agreement)
- Services your child will receive when and where they will receive them; and who will deliver the services
- Steps to be taken to support his or her transition to another program

The IFSP may also identify services the family may be eligible for, including financial information or information about raising a child with a disability.

### Early childhood transition

At least 90 calendar days (or up to six months depending on the State) prior to a toddler reaching the age of 3, the responsible agency\* of Early Intervention services must notify the school district where the child lives that the child will be eligible for preschool services under IDEA 2004. Under IDEA the lead agency must ensure a smooth transition and evaluation of each child eligible for school age services under IDEA 2004.

*\*Each State is different regarding which public agency is responsible for the implementation of early childhood under Parent C of IDEA 2004. (Health Department, DD (Developmental Disability) Council, School District).*

## Section Five Appendix

Organization	Description	Website
<b>CDC: Early Intervention</b>	Early Intervention contact information by state.	<a href="http://www.cdc.gov/ncbddd/actearly/parents/state-text.html">www.cdc.gov/ncbddd/actearly/parents/state-text.html</a>
<b>Zero to Three</b>	A national, nonprofit organization that provides resources in the first three years of life. Their goal is to strengthen and support families, practitioners, and communities to promote the healthy development of babies and toddlers.	<a href="http://www.zerotothree.org">www.zerotothree.org</a>
<b>Teaching Strategies</b>	This publishing and training company's mission is to offer high quality early childhood curriculum materials, training materials and parenting resources.	<a href="http://www.teachingstrategies.com">www.teachingstrategies.com</a>
<b>The Division for Early Childhood (DEC) of the Council for Exceptional Children (CEC)</b>	Promotes policies and advances evidence-based practices that support families and enhance the optimal development of young children (0-8) who have or are at risk for developmental delays and disabilities.	<a href="http://www.dec-sped.org">www.dec-sped.org</a>
<b>National Association for the Education of Young Children</b>	The nation's largest organization of professionals dedicated to improving the quality of early childhood education programs for children birth through age 8. Website includes a section for parents.	<a href="http://www.naeyc.org">www.naeyc.org</a>

## Section Six: Educational rights

### Services under IDEA

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

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

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

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

### School evaluations

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

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

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

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

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

All testing or evaluations must:

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

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

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

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

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

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

## **Developing an Individual Education Program (IEP)**

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

## **Preparing for an IEP meeting**

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

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

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

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

Members of an IEP Team should include the following:

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

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

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

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

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

## Understanding IEP components

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

## IEP requirements

The IEP must include the following information:

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

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

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

## Measurable annual goals

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

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

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

## Additional services in the school setting

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

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

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

**Therapy**  
Speech

**How Often**  
2x weekly 20 min

**Who**  
Speech Therapist

**Where**  
Small Group/Resource Room

## Section Six Appendix

Organization	Description	Website
<b>Department of Education</b>	Our mission is to promote student achievement and preparation for global competitiveness by fostering educational excellence and ensuring equal access.	www.ed.gov
<b>Individuals with Disabilities Education Act (IDEA)</b>	Access from the U.S Department of Education website	www.sites.ed.gov/idea/about-idea/
<b>Office of Civil Rights (OCR)</b>	Access from the U.S Department of Education website	www.ed.gov/about/offices/list/ocr/aboutocr.html
<b>Center for Parent Information &amp; Resources</b> <i>Available in English and Spanish</i>	<p><i>Notable articles:</i></p> <p><i>“Developing Your Child’s IEP”</i></p> <p><i>“When the IEP Team Meets”</i></p> <p><i>“Questions Often Asked by Parents about Special Education Services”</i></p> <p><i>“Contents of the IEP”</i></p> <p><i>“Special Factors in IEP Development”</i></p>	www.parentcenterhub.org
<b>Understood for All Inc.</b> <i>Available in English and Spanish</i>	<p>Sample letter requesting evaluations and reports for services.</p> <p><i>Notable Articles:</i></p> <p><i>“What is Early Intervention?”</i></p> <p><i>“The difference between IEPs and 504 plan”</i></p> <p><i>“What is Special Education?”</i></p> <p><i>“4 Benefits of Inclusive Classrooms”</i></p> <p><i>“What is the Individuals with Disabilities Education Act (IDEA)?”</i></p> <p><i>“The 13 disability categories under IDEA”</i></p>	www.understood.org



## IEP Meeting Information:

Date: 3/14/21

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

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

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

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

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

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

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

## Notes:

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

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

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

## Section 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.

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

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

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

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

Assistive technologies can include mechanical, electronic, and microprocessor-based equipment, non-medical and non-electronic aids, specialized instructional materials, services and strategies 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. In many cases, where an IEP includes specific requirements for these tools, the school system will be responsible for the cost. Outside of IEPs, funding can come from government programs (SSI, Medicaid, etc.), private insurance, employers or local charities or disability support organizations. For additional information, it is recommended to visit the Assistive Technology program in your state to explore what funding or support is offered in your area.

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

## Section Seven Appendix

Organization	Description	Website
<b>Seizure Tracker</b>	Founded by parents of a child with TSC who were struggling to understand their son’s seizure activity, Seizure Tracker™ has become a valuable tool for parents, doctors and researchers alike.	<a href="http://www.seizuretracker.com">www.seizuretracker.com</a>
<b>KidNeeds</b>	Provides resources for special needs children and their families for different types of disabilities, growth, development, and technology.	<a href="http://www.kidneeds.com">www.kidneeds.com</a>
<b>Danny Did Foundation</b>	A resource for families who are impacted by seizures  <i>Notable Section: Devices + Technology</i>	<a href="http://www.dannydid.org">www.dannydid.org</a>
<b>Epilepsy Foundation</b>	To lead the fight to overcome the challenges of living with epilepsy and to accelerate therapies to stop seizures, find cures, and save lives.  <i>Notable Section: Epilepsy Device Wiki</i>	<a href="http://www.epilepsy.com">www.epilepsy.com</a>
<b>Charlie Foundation</b>	Dedicated to advancing the awareness of ketogenic therapeutics to help with seizures.	<a href="http://www.charliefoundation.org">www.charliefoundation.org</a>
<b>Chelsea Hutchison</b>	A non-profit to provide help and support to individuals, particularly children and young adults, who have epilepsy.  <i>Notable Section: Helpful Links: Monitoring Devices</i>	<a href="http://www.chelseahutchisonfoundation.org">www.chelseahutchisonfoundation.org</a>

## Section Eight: Self care and family dynamics

### Caregiver mental health

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

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

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

In the midst of being overwhelmed, it can also be challenging to even know where to start, how to recognize if you are close to “burning out,” and what action steps you can take to create healthy boundaries so you can live your fullest life. Below are some signs and symptoms of caregiver burnout. If you or another caregiver is experiencing these symptoms, we recommend reaching out to your local provider or therapist to work through your unique situation and develop a 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 hear, “how can I help you.” Yet, you might not even know that because you are already in a crisis and decision fatigue is already setting in. Having a go to list that you can show to others not only helps you but helps those in your inner circle to know how to truly help you.

Locate the Finding Your Caregiver Zone and How to Help Me guide and worksheet in the appendix of this section for additional support. Please visit the TSC Navigator periodically for added resources and updated content.

### Signs and symptoms of caregiver burnout

If you find yourself identifying with a lot of these feelings, please reach out to your primary care physician or local mental health provider.

#### Physical

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

## Emotional

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

## Behavioral

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

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

## 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 that everyone on this journey has experienced grief in their own unique way. While it's of course natural for parents to focus primarily on the wellbeing of their child, it's important to recognize the life you once had will forever change. Your routines, connections and friendships may change – of course, this is a common part of parenthood even outside the realm of a rare disease diagnosis. 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, lifechanging moments, it is important to shed 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 that families and individuals thrive when given unconditional support. So, let's start with where you are.

The utmost important factor to remember is you will cycle through a continuum of grief. 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. Some of these 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 to put a timeline on when you should be “over it” or belittling statements of 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 friendship in place to help be a guardrail for these signs and symptoms.

It is also okay to have your inner circle of friends educated on 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 nonjudgmental presence might be all that 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 still 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 a relationship with your child with TSC, help them be authentic. Teach them the best ways to engage your child. 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.

## Family discussions

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

## How to talk to your child about TSC

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

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

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

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

The most vital thing you can do is give your unaffected child(ren) consistent individual time with them. Let them express their feelings for how they feel and not tell them how they feel. It's okay if their emotions do not match with the reality of the situation, but with consistent individual time together, improved coping and communication skills will develop. It is also important to set a healthy example and verbalize it's okay to ask for help. It's okay for things to feel like they are not okay and verbalizing that is important. Another key factor is explaining the disability to your unaffected child(ren) at their age-appropriate level of understanding, honestly as possible. It is essential to meet your unaffected child(ren) where they are at each milestone, emotion and significant event that occurs throughout this journey, which like your affected child, will change over the years. Providing peer support and professional assistance can also help your unaffected child(ren) develop the skills as they too are on this journey. Another helpful option is to help your unaffected child(ren) articulate what to say when things might come up in public or at school. Helping them proactively have problem solving skills can help eliminate barriers and issues on their TSC journey.

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

## Respite care

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

To find out if there is an affiliate in your community that provides this service, visit The Epilepsy Foundation at [www.epilepsy.com](http://www.epilepsy.com). In addition, the National Respite Locator Service helps individuals, parents and caregivers find respite services in their state and local area. To learn more about types of respite care please visit TSC Navigator: ABCs of Respite: A Consumer Guide for Family Caregivers.

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

## Section Eight Appendix

### General family resources

Organization	Description	Website
<b>Child Neurology Foundation</b>	To serve as a collaborative center of education, resources and support for children and their families living with neurologic conditions and facilitate connection with medical professionals who care for them.  <i>Notable Articles: Tools &amp; Resources Preparing for Your Doctor's Visit Support for Patients &amp; Caregivers</i>	<a href="http://www.childneurologyfoundation.org">www.childneurologyfoundation.org</a>
<b>Charlie Foundation</b>	Was founded in 1994 to provide information about diet therapies for people with epilepsy, other neurological disorders and select cancers.  <i>Notable Articles: Ketogenic Therapies &amp; Resources</i>	<a href="http://www.charliefoundation.org">www.charliefoundation.org</a>
<b>Variety</b>	Variety-The Children's Charity serves children who may fall through the cracks of government funding or other aid. Depending on government policy and the availability of other assistance, items and services granted by Variety do vary from state to state.	<a href="http://www.usvariety.org/about-us/">www.usvariety.org/about-us/</a>
<b>Special Needs Project</b>	Provides books about disabilities, that serve individuals, families, and professionals with a large collection of disability-related materials, including titles about Autism, ADHD, Independent Living and Full Inclusion.	<a href="http://www.specialneeds.com">www.specialneeds.com</a>

<b>The M.O.R.G.A.N. Project</b>	The M.O.R.G.A.N. Project focuses on awareness and support of families and parents caring for special-needs children. It provides families with specialized programs for children with special health care needs, disabilities, sensory issues and concerns to improve family quality of life.	<a href="http://www.themorganproject.org">www.themorganproject.org</a>
---------------------------------	---	--

## Parent and caregiver resources

Organization	Description	Website
<b>Courageous Parents Network</b> <i>Languages available: English, Spanish</i>	Empowers, supports and equips families and providers caring for children with serious illnesses.	<a href="http://www.courageousparentsnetwork.org">www.courageousparentsnetwork.org</a>
<b>Parents Helping Parents (PHP)</b>	Supports, educates and inspires families and the community to build bright futures for youth and adults with special needs.	<a href="http://www.php.com">www.php.com</a>
<b>Association for Successful Parenting</b>	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.	<a href="http://www.achancetoparent.net/">www.achancetoparent.net/</a>
<b>The National Alliance for Caregiving (NAC)</b>	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.	<a href="http://www.caregiving.org">www.caregiving.org</a>
<b>Parent to Parent USA</b>	Provides emotional and informational support to families of children who have special needs.	<a href="http://www.p2pusa.org">www.p2pusa.org</a>

## Sibling resources

Organization	Description	Website
<b>Sibling Leadership Network</b>	The purpose of the Sibling Leadership Network is to promote a broad network of siblings who share the experience of disability and people concerned with sibling issues by connecting them to social, emotional, governmental, and provisional supports across the lifespan enabling them to be effective advocates with their siblings and to serve as change agents for themselves and their families.	<a href="http://www.siblingleadership.org">www.siblingleadership.org</a>

<b>The Sibling Support Project</b>	A national program dedicated to the interests of siblings of people with disabilities. Provides peer support and educational opportunities for siblings of individuals with disabilities.	<a href="http://www.siblingsupport.org">www.siblingsupport.org</a>
<b>Siblings with a Mission</b>	Serves and supports siblings and families of individuals with complex health conditions and developmental disabilities.	<a href="http://www.siblingswithamission.org">www.siblingswithamission.org</a>

## Mental health resources

Organization	Description	Website
<b>The National Federation of Families for Children's Mental Health</b>	Provides advocacy for the rights of children and youth with emotional, behavioral, and mental health challenges; provides assistance to a network of family run organizations; and collaborates with organizations to transform mental health care in America.	<a href="http://www.ffcmh.org">www.ffcmh.org</a>
<b>Mental Health America (MHA)</b>	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.  <i>Notable Article:</i> <i>Mental Health Resources for Caregivers</i>	<a href="http://www.mhanational.org">www.mhanational.org</a>

## Respite services

Organization	Description	Website
<b>David's Refuge</b>	Based out of New York, David's Refuge promotes "caring for the caregiver" and hosts weekend B&B getaways for parents and caregivers of children with special needs. It focuses on families in the New York area, but if you live anywhere on the east coast and can drive to New York for one of these respite weekends, it will extend services to you!	<a href="http://www.davidsrefuge.org">www.davidsrefuge.org</a>
<b>ARCH National Respite Network</b>	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.	<a href="http://www.archrespite.org">www.archrespite.org</a>
<b>A Mother's Rest</b>	Is a wonderful organization that provides discounted caregiver/respite retreat weekends across the US. Its mission is to improve the emotional and physical health of caregivers through proactive, restorative, respite opportunities.	<a href="http://www.amothersrest.org">www.amothersrest.org</a>



## Finding Your Caregiver Zone

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

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

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

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.?



8737 Colesville Road, Suite 400 | Silver Spring, MD 20910  
800-225-6872 | [info@tscalliance.org](mailto:info@tscalliance.org) | [tscalliance.org](http://tscalliance.org)

Dear Family Member:

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

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

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

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

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

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

**Hope no matter how complex**



8737 Colesville Road, Suite 400 | Silver Spring, MD 20910  
800-225-6872 | [info@tscalliance.org](mailto:info@tscalliance.org) | [tscalliance.org](http://tscalliance.org)

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

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

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

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

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

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

Sincerely,

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

**Hope no matter how complex**





**Hope no matter how complex™**

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