

## Tuberous Sclerosis *At a Glance*

Tuberous Sclerosis Complex (TSC) is a highly variable condition that includes growth of many (non-cancerous) tumors in the body. The tumors can occur in the heart, skin, brain, kidneys, and other organs. They can sometimes lead to serious health problems. TSC can also cause learning and behavioral problems.

This condition varies even within a family. It can range from a mild disorder affecting the skin and brain; to epilepsy, intellectual disability, autism spectrum disorder, and kidney disease.

- TSC is a dominant inherited genetic disorder, occurring in about 1 in 5,800 births. Most cases occur due to a spontaneous mutation.
- It is also known as Tuberous Sclerosis 1 (TSC1), Tuberous Sclerosis 2 (TSC2), and Bourneville Disease. **A blood test may be able to determine the exact gene change in either the TSC1 gene or the TSC2 gene.**

### *Common features of Tuberous Sclerosis Complex:*

#### Skin issues

- Almost everyone with TSC has skin issues. These may include various spots and raised patches on the skin, side of nose and on cheeks, forehead, lower back, and around the nails. The skin problems do not result in serious medical problems; however, they can cause problems with self-esteem as the child gets older. The facial angiofibromas usually start developing around the age of 5.
- There may also be white (unpigmented) spots on the skin that can be detected with a special light (Wood's lamp)--as well as locks of hair that have no pigment.

#### Brain issues

- Characteristic brain issues include cortical brain tubers and subependymal nodules.
- Cortical brain tubers can be found in different parts of the brain and may act as a seizure focus.
- Subependymal nodules occur in 90% of people with TSC and most remain dormant throughout life, but can cause problems if they obstruct the flow of the fluid around the brain.

- Subependymal nodules develop into subependymal giant cell astrocytomas (SEGA) in 6-14% of people with TSC and are one of the leading causes of medical problems and death.
- Cortical brain tubers and subependymal nodules are thought to cause seizures, or affect learning, mood, or behavior.
- Seizures occur in 60-90% of people who have TSC. Young children may have infantile spasms which are clusters of seizures that resemble a startle reflex. Medications can help treat these seizures and help reduce impact on function.

### Kidney issues

- About 80% of children with TSC have a renal lesion by 10.5 years.
- Kidney growths are common in people with TSC.
  - These growths can cause problems with kidney function. They may be life threatening in some cases.
  - An increase in blood pressure, back pain, or blood in urine can be a sign of kidney tumor growth.

### Heart issues

- Benign tumors (known as rhabdomyomas) that are usually present at birth and do not cause a problem unless they are in a spot that obstructs the flow of blood or causes a problem with the pacemaker. These benign tumors do not grow and may become smaller over time and may not be detectable by ultrasound as an adult.
- Arrhythmias (irregular heartbeats) are usually present early in life and are treated. Rarely, the irregular heartbeat may persist.

### Lung issues

- Tumors may occur in lungs (Lymphangiomyomatosis (LAM)). Mean age of diagnosis of LAM is 28 years. However, these can occur in teenagers.
- More common in females than males (40% of woman have LAM)
- Signs:
  - Shortness of breath after mild exercise
  - Cough
  - Lung collapse

### Eye issues

Benign eye tumors (Hamaratomas)

- Don't usually cause symptoms
- Don't typically cause visual loss or problems.

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## Teeth issues

- Gum tumors and dental pits can be extensive in the adult teeth.

## Intellectual disability / developmental delay

- Intellectual disability/developmental delay occurs in roughly 50% of individuals with TSC.
- Early diagnosis and intervention is key.

## Cognitive, behavioral, and psychiatric issues

- Attention deficit hyperactivity disorder (ADHD)
- Behavioral and psychiatric disorders, often part of the autism spectrum disorders (ASD)
  - About 25- 60% are diagnosed with ASD

## ***Things to Think About***

### 1. Medical/Dietary Needs

#### What you need to know

The list of possible medical problems in TSC is quite extensive. However, each individual usually has only some of these problems. Also, the severity of any one of these medical problems varies widely in individuals. Therefore, it is important to ask the parents about the medical issues in their child.

School age children with TSC should have annual doctor and specialist visits to monitor medical conditions. A TS clinic can offer many specialists under one roof.

#### *Learn more*

#### Recommended:

- Annual physical
- Annual eye exam
- Regular developmental assessment of children
- Watch for and treat seizures:
  - Early control of seizures can reduce behavioral consequences.
    - Seizures can be very subtle or unnoticeable.

No special diet is required for TSC, although a well-balanced diet is important. Some children may be on a ketogenic diet to help with seizure control.

## What you can do

- A yearly checkup and studies as needed should occur in the child's Medical Home – their primary care office where care can be coordinated.
- Notify the parents of changes in child's energy level or if there is pain.
- Be aware of changes in behavior or mood that aren't typical. Notify the parents.
- Be aware of academic, social, behavioral changes. Contact parents when changes are noticed.
- Be an advocate in obtaining sufficient speech and language support during school to communicate effectively throughout the day. This may include augmentative communication devices.
- Monitor child for possible seizures.
  - Seizures can be very subtle or unnoticeable.
    - For example, a student may stare for just seconds.

## 2. Education Supports

### What you need to know

See *TSA Teacher's guide: Educating a child with TSC*

<http://www.tsalliance.org/documents/Teacher%20Guide%20to%20TSC.pdf>

Although some children with TSC have significant problems such as epilepsy and significant learning disabilities, *many children have no symptoms in the classroom*. The condition may impact the child's ability to learn, his/her behavior, and relationships. **It is important to have HIGH EXPECTATIONS for learning** for children who have TSC. TSC experts recommend that children with TSC have a thorough neuropsychological evaluation at time of diagnosis so early intervention can start.

Neuropsychological challenges often present in writing, reading, spelling, and arithmetic. Children often work well with one-on-one assistance in highly structured environments. Tasks can be broken into simple steps and instructions repeated over short periods. Behavior and learning issues may be a red flag for changes in tumor growth and seizure activity in the brain.

In adolescence, the following may be appropriate:

- A vocational assessment noting cognitive strengths and challenges
- Teaching adaptive behavior and daily living skills.

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## Learning difficulties

Executive control challenges:

- Executive control skills are needed for purposeful, goal-oriented activity. They play a role in a child's intellectual development, academic achievement, personality, social skills, relationships, and communication with others.
- May have difficulty with working memory, planning, organization, and complex problem solving.
- These difficulties affect all areas of learning.
- Some differences are subtle.
- Because of planning and organization difficulties, starting an action may be challenging.
- Children may be overwhelmed by projects.
- They may be described as inflexible and concrete thinkers.

Learning, attention, and memory challenges:

- They may have problems sustaining attention and switching tasks
- Abstract language and concepts may be difficult to grasp
- They may not understand or use metaphors.
- Instructions may be heard but not retained long enough for action
- May have difficulty with integration skills, working memory, and episodic memory problems

## Reading challenges (dyslexia)

Early signs may include difficulty with:

- Moving to the rhythm of music
- Remembering content of stories
- Understanding left to right
- Understanding visual spatial concepts
- Coordination (skipping is hard)
- Using correct words or reversing words when speaking
- Writing letters, words, and/or numbers (reversals)
- Proof reading their own written work
- Understanding time
- Understanding seasons

## Math challenges (dyscalculia)

The inability to understand the meaning of numbers or use math principles to solve problems is called **dyscalculia**. When a child has TSC involvement in the language and visual process centers of the brain, dyscalculia can occur.

Early signs may be difficulty with any of the following:

- Understanding numbers and quantities
- Understanding addition, subtraction, multiplication, division, abstract concepts (algebra)
- Making change
- Handling money
- Recognizing patterns
- Understanding days, weeks, months, and time
- Lining up numbers on page
- Telling time

### Challenges with writing and processing spoken language (dysgraphia)

A learning disability that affects written expression and difficulty in processing spoken language is called **dysgraphia**. Individuals with TSC have visual-spatial difficulties and struggle to organize letters, numbers, and words on a line or page. They may also have language processing difficulties.

Early signs may be difficulty with:

- Writing or forming letter shapes
- Being consistent with spacing between words and letters
- Stamina when writing even short assignments
- Writing assignments (refusals, reluctance)
- Drawing or coloring
- Holding pencil grip or using appropriate pressure on pencil point
- Staying on lines when cutting with scissors
- Getting thoughts on paper (older students)

### Communication

- There are individuals with TS who have no intellectual or communication challenges.
- Receptive language is usually good. However, some individuals have difficulty using interactive language for social communication.
- Word retrieval challenges can cause problems with expressive language.

### What you can do

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## Interventions for communication challenges

- Individuals with speech and motor issues often benefit from speech and occupational therapy.
- Promote language understanding by using simple short sentences, visual prompts, and pictures.
- Use a child's experiences and interests to engage child in learning.
- Allow extra time, repeat directions, provide lesson summaries, and/or record lesson so child can listen again.
- To promote language development:
  - Provide ample time for responding
  - Increase self-confidence by calling on them when they know answer
  - Encourage child to repeat the questions before responding
  - Allow a child time to rehearse and respond

## Interventions for attention and memory

- Help with organization.
- Present information in concrete manner.
- Use manipulatives to show concepts.
- Simplify verbal information.
- Explain concepts clearly.
- Provide visual cues and instruction.
- Repeat information.
- Use positive reinforcement.
- Ask child to repeat instructions. Help the child find a starting point, especially on complex tasks. They often have a hard time with multiple step tasks and lose track of what they are doing.
- Select relevant task goals.
- Use a calendar to track important events.
- Help them develop a means to solve complex problems.
- Monitor and evaluate behavior and emotions.
- Help organize common needs at school and at home. For example, have a place for all things, use different colored notebooks for different subjects, etc.

## Interventions for visual spatial problems and motor skills

- Modify copying. For example, provide a copy of teacher's or other student's notes.
- Provide a simple overview or summary before lessons.
- Provide clear tests that are as simple as possible with only a few problems on a page. Graph paper may help, especially in math.
- Use lined paper to help place responses.

- Allow extra time on work.
- Practice tracing shapes and copying pictures.
- Provide feedback to help awareness of mistakes.
- Use verbal descriptions to reinforce visual information.
- Practice folding and cutting with scissors.
- May have difficulty matching shapes and sizes. Puzzles may be challenging.

### **Interventions for motor and sensory development**

- Occupational and physical therapy may be helpful.
- Visual instruction may work better than verbal.
- Limit the amount of written homework.
- Use manipulatives (Legos, play dough).
- Practice cutting.
- Allow longer time to write.
- Write on every other line.
- Allow tracing.

### **Interventions for reading challenges (dyslexia)**

- Early identification and intervention
- RTI (response to intervention) program

### **Interventions for Math challenges (dyscalculia)**

- Assistive technology (calculator)
- Use music to teach math facts
- Use pictures for word problems
- Color code math problems (red= subtraction, green= addition)
- Use manipulatives or number charts
- Use graph paper to keep numbers in line
- Assess curriculum to determine what part(s) will help the student reach their potential.
- Students often can learn basic math concepts (telling time, counting money, etc.) but may have difficulty learning algebra and geometry.

### **Interventions for writing and processing spoken language (dysgraphia)**

- Use computer for writing assignments
- Provide occupational therapy
- Use a tape recorder to record ideas and organize thoughts
- Have a peer take notes
- Test orally

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- Minimize writing assignments
- Do not have students “slow down.” This causes them to get stuck on the details for writing and they may lose their train of thought

Information comes from the Children’s Tumor Foundation [www.ctf.org/](http://www.ctf.org/) Please see their website for additional information.

### 3. Behavioral and Sensory Supports

#### What you need to know

See Tuberous Sclerosis Alliance: Behavioral issues and TSC

<http://www.tsalliance.org/documents/Behavioral%20Issues%20and%20TSC.pdf>

Most individuals with TSC usually exhibit at least one mental health issue. Many children with TSC have behavioral issues.

#### Autism Spectrum Disorders (ASD)

- Autism (ASD) is diagnosed in about 25-60% of children with TSC. TSC is considered one of the leading genetic causes of ASD.
- There is a wide range of abilities in this diagnosis.
  - Individuals with very significant global intellectual disabilities have ASD up to 50% of the time.
  - Social communication difficulties are found in 44-69% of individuals with TSC.
  - Some individuals with ASD may have problems fitting into school and life.
- Findings include
  - Poor eye contact
  - Repetitive and ritualistic behaviors
  - Speech and language delays
- Treatment options vary based on individual’s age and ability.
  - Foster skills in the three main areas of difficulty:
    - Social skills
    - Communication skills
    - Imaginative play
  - Ensure repetitive or obsessive behaviors don’t become too prominent or interfere with family life.
  - Treatment aims to help parents support their child’s development.

#### Mood related difficulties

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### **Anxiety:**

- Individuals with TSC have a high rate of anxiety.
- They may have a co-existing mental health condition, such as depression.
- In older children, anxiety and mood related disorders are more common.
  - May appear as excessive worry, unexplained panic attacks, acting out.

For more information see *“What are Anxiety Disorders” and WorryWiseKids.org.*

- <http://www.nimh.nih.gov/health/topics/anxiety-disorders/index.shtml>
- <http://www.worrywisekids.org/node/52>

### **Aggressiveness/Disruptive behaviors (40-50%):**

- Can have outbursts and behaviors that cause self-injury.
- Some medications can cause aggressive behaviors.
- May appear as increased activity, restlessness, impulsivity, aggressive outbursts, temper tantrums, and self-injurious behaviors.

### **Depression:**

- Depression often occurs with other mental health issues like anxiety.

For more information see *“What is Depression?”*

<https://www.nimh.nih.gov/health/topics/depression/index.shtml>

### **OCD:**

- May have rituals and repetitive behavior

For more information see *“Obsessive-Compulsive Disorder Signs & Symptoms”*

<http://www.nimh.nih.gov/health/topics/obsessive-compulsive-disorder-ocd/index.shtml>

### **ADHD:**

- As many as 25-50% of children TSC have some signs of ADHD.

For more information see *“What is Attention Deficit Hyperactivity Disorder (ADHD, ADD)?”*

<http://www.nimh.nih.gov/health/topics/attention-deficit-hyperactivity-disorder-adhd/index.shtml>

## **What you can do**

### **Strategies for children who have autism**

- Provide information and discuss similarities and challenges as a class.
- Help develop confidence and focus on strengths.
- Provide positive reinforcement.

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- Be consistent with directions, rules, and discipline. Ensure they have positive behavioral supports.
- Teach child appropriate social behaviors/skills (role model, friend groups). Help them nurture friendships.
- Teach how to recognize facial expressions, body language, and moods in self and others.
- Ensure effective communication systems and AAC devices are available if needed.
- Ensure the right people are trained in using the AAC device, including the child.
- Ensure needed supports for success in school.
- Ensure the team has training, support, meeting time.
- Ensure interventions match the child's unique needs.

### Strategies for mood disorders

- Early identification and behavioral interventions
- Unstructured situations may be difficult for individuals with mood and anxiety disorder (e.g., school assemblies, in-between classes, on bus, and on field trips).
  - Watch for overload.
  - Prepare them ahead of time, offer supports, redirect behaviors.

### Strategies for ADHD

- Seat student where distractions are minimal
- Post schedule and assignments in easy viewing
- Review daily schedule visually *and* verbally
- Give reminders of schedule throughout the day
- Review homework schedule
- Prepare for transitions
- Let student know when transitions are going to occur (alerts at 15, 10, and 5 minutes before they occur)
- Help with daily organization
- Pair student up with well-organized peer
- When giving instructions, provide a visual example of the instructions to ensure understanding
- Break instructions into steps
- Provide priority lists for large assignments
- May need extra time for test
- May need small manipulative to help focus when listening
- Use communication book between teachers and parents
- Develop rules and routines

## 4. Physical Activity, Trips, Events

No special accommodations are needed for individuals with TSC, unless required due to the child's learning and/or behavioral difficulties. May need emergency medication in case of seizure.

- May need extra preparation about what to expect during the activity
- See additional supports in the Educational Interventions section.

Individuals who have TSC are not usually limited in their activities. The only exception is for those children with a specific complication, like a heart arrhythmia.

- A medical doctor will point out if there are any restrictions.

If you live in New England (USA) and qualify, Northeast Passage offers Therapeutic Recreation and Adaptive Sports programming ([www.nepassage.org](http://www.nepassage.org)).

## 5. School Absences and Fatigue:

### What you need to know

- The most common sleep problem is insufficient sleep which affects both those with TSC and their caregivers.
- About 20-50% of individuals with TSC have sleep problems.
  - daytime sleepiness
  - going to sleep
  - staying asleep
  - underlying sleep disorders
- Sleepiness may manifest itself as hyperactivity in children and they will fight the sense of sleepiness by increasing motor activity to stay awake.
- Epilepsy and sleep issues:
  - seizures disrupt sleep and lack of sleep increases seizures
  - anticonvulsant medications can cause daytime sleepiness
  - insufficient sleep is the primary cause of daytime sleepiness
- School aged children who have TSC may have increased school absences due to medical issues.

## 6. Emergency Planning

### What you need to know

- Emergency plans are individually determined, based on child's behaviors and medical issues.

### What you can do

- Mention any new signs, symptoms, and/or pain to the child's parents.
- These sample forms <http://opi.mt.gov/pdf/health/healthcareneeds.pdf> from the Montana Office of Public Instruction include a section on emergency procedures which may be helpful, especially for parents.
  - Just one example; you will find others

## 7. Resources

### Tuberous Sclerosis Alliance

<http://www.tsalliance.org>

*The TSA was founded in 1974 by four mothers coming together to provide fellowship, generate awareness, pursue more knowledge and provide hope to those that shared the common bond of tuberous sclerosis complex. These goals are still driving the organization today.*

This website includes information for schools as well as for families, health professionals and researchers. <http://www.tsalliance.org/pages.aspx?content=567>

Tuberous Sclerosis Alliance: Behavioral Issues and TSC

<http://www.tsalliance.org/documents/Behavioral%20Issues%20and%20TSC.pdf>

### Medline Plus

<http://www.nlm.nih.gov/medlineplus/tuberoussclerosis.html>

Includes a range of information on TSC, including overview, disease management, clinical trials

### Genetics Home Reference

<https://ghr.nlm.nih.gov/condition/tuberous-sclerosis-complex>

Consumer-friendly information about human genetics from the U.S. National Library of Medicine

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## Meet a Child with TSC – *Sophia’s Story*

*GEMSS would like to thank Sophia and her mother for their generosity in sharing this story with us. You have made the site come to life with the addition of your thoughts and feelings. Thank you so much!*

Sophia, age 6, is having an “incredible” year at school states her mother, Elisa. She is in a regular classroom and has a paraprofessional supporting her. After hearing that she may never speak, they are delighted to hear her speak with clear words, using 3-4 words in full sentences. At school and home, she uses an iPad which helps with spelling and practicing writing. Sophia is quite social and seeks out certain people. She is included in regular class activities and it works well when “everyone works together.” Elisa feels that Sophia’s team is so invested and genuinely interested in helping Sophia reach her full potential.



Sophia was diagnosed with Tuberous Sclerosis at sixteen months of age.

Sometimes, Sophia gets frustrated when things don’t go as planned. She has behaviors that might frighten other children, such as screaming when she is upset. This particularly may happen after a seizure. Her behavior can make it harder to get out in the community. As more supports are put in place, life gets easier for Sophia and those around her. For example, a behavior specialist has been visiting with Sophia’s family to teach everyone how to deal with the negative behaviors, how to keep people safe and how to provide strategies to avoid or lessen these occurrences in the future. A weighted vest at school has also been a good sensory support for her.

Sophia lives with her sister and parents in the United States in a rural area near the Canadian border. Family is not very close by but the family gets supports through Respite which “is a Godsend.” Sophia spends “Tuesdays with Terry” and Terry’s family has developed an extended family in their household. Family Support has also been excellent in helping figure out funding issues. Sophia’s sleep issues make it difficult for the family at times. Her behaviors used to be more self-directed, however, have become more outwardly directed over time. The family has created a great play space for Sophia so that she can have a quiet place when she needs it. Elisa’s advice for other parents and teachers is to learn about TS and to be open to suggestions and discussion. “Being in regular classes has done a world of good for her.” Sophia gets to see

how her peers act socially in many different environments. Doing this has enabled Sophia to grow in many areas personally and socially.

There is a Seizure Action Plan in place which is supported by her medical community. Elisa advises school staff to talk through changes, to talk through new ideas, and to talk through proposed strategies with parents. For example, having Sophia carry a big backpack full of supplies on an outing really would stigmatize her as no one else would have to carry a pack. Talking it through helped the team come up with acceptable alternatives.

Being a parent to a child with Tuberous Sclerosis, Elisa has learned something very valuable she would like to share. “Don’t think you know their potential. They can surprise you!”

**Note:** *This printable version does not include the information found under the green button marked “Transitions” on the website. Those general pages may be printed separately.*