

Understanding seizures with tuberous sclerosis complex (TSC)



Maddie, at age 7
Living with TSC



Seizures, TSC, and the journey ahead

YOU ARE
NOT ALONE

APPROXIMATELY
1 MILLION
PEOPLE
WORLDWIDE ARE
CURRENTLY LIVING
WITH TSC

INCLUDING UP TO
50,000
PEOPLE
IN THE US

TSC is a term that you may have never heard before you or your loved one received a diagnosis—and, since then, it's all you seem to think about.

Whether you or your loved one just received a diagnosis, or have been living with seizures and TSC for some time, a million thoughts are probably running through your mind.

While the idea of seizures associated with TSC may bring a lot of fear and questions, you can take valuable action by learning how to work with your family and care team to prepare for seizures, identify them, and even reduce their occurrence.

We hope you'll find comfort in knowing there is a great deal of research, support, and guidance available to you.



Jack, at age 11
Living with TSC

BRAVE
STRONG
FEARLESS
WARRIOR





Alex, at age 21
Living with TSC



What is TSC?

Tuberous sclerosis complex (noun): too-ber-uh-s, skli-roh-sis, kom-pleks—a highly variable genetic disorder that is most likely not inherited, but can be passed on from the person’s parent(s). TSC causes noncancerous tumors to develop in many different organs, including the brain, skin, kidneys, lungs, heart, and eyes. TSC affects each person differently, with signs and symptoms ranging from very mild to severe.

Possible signs and symptoms of TSC

TSC affects different organs in the body. The following table contains some of the signs and symptoms that may occur in TSC. *You or your loved one may not experience all of these.*



BRAIN

- Seizures
- Noncancerous structures on the outer and/or inner layers of the brain
- Behavioral disorders and intellectual disabilities



SKIN

- Small red bumps on the face
- Small fibrous growths on fingernails and/or toenails
- Plaques on the forehead or scalp
- Tough patches of skin
- White spots on the skin



HEART

- Noncancerous cardiac tumors



KIDNEY

- Noncancerous tumor structures
- Multiple cysts
- Polycystic kidney disease



LUNG

Women

- Formation of cysts in the lungs that may or may not cause symptoms

Men and women

- Overgrowth of cells in the lining of the air sacs

OTHER

- Noncancerous structures affecting the tongue, gums, or eyes

How common are seizures in TSC?

Emily, at age 22
Living with TSC

ABOUT
85%
EXPERIENCE
SEIZURES THROUGHOUT
THEIR LIFETIME



NEARLY
100%
DEVELOP RECURRING
SEIZURES AFTER HAVING
THEIR FIRST SEIZURE

Seizures are episodes of disrupted brain function that cause changes in attention, behavior, and/or muscle control, and can cause a significant burden in TSC.

Seizures are the most common neurological symptom of TSC

While the reason for seizures is not definitively known, many people with TSC have abnormal structures in the brain that are thought to cause seizures.

Some types of seizures are more common than others

Most people with TSC can experience different types of seizures; however, infantile spasms and focal seizures, also known as partial seizures, are the most common.

1 IN 3

INFANTS DEVELOP
INFANTILE SPASMS

UP TO

82%

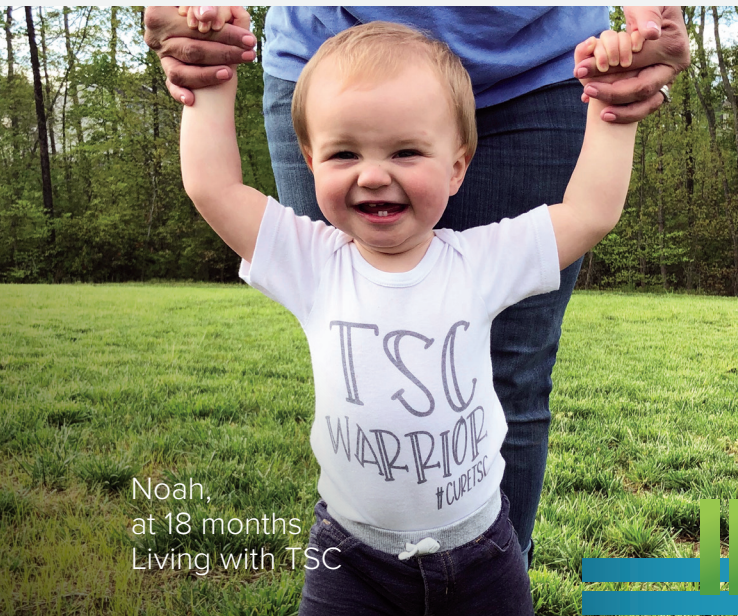
OF PEOPLE EXPERIENCE
FOCAL SEIZURES

SEIZURES CAN RANGE
FROM VERY

MILD TO
SEVERE



The importance of early recognition, diagnosis, and treatment



Noah,
at 18 months
Living with TSC

Unrecognized and/or uncontrolled seizures are linked to developmental disabilities, learning and behavioral disorders, and other negative long-term outcomes.

By catching seizures early on, you and your doctor can develop the right treatment plan.



Uncontrolled or undetected seizures can **increase** the severity of



**Risk of injury
due to falls**



**Learning
and/or cognitive
impairments**



**Developmental
disabilities**

A few tips to help:

Talk with your doctor as soon as you notice a seizure in you or your loved one.

According to the Epilepsy Foundation, **seek emergency medical attention** if a seizure lasts for more than 5 minutes or there is no recovery in between seizures.

As difficult as it may seem in the moment, **take a video** to show your doctor if you suspect you or your loved one is having a seizure.

The information provided is not intended to replace a doctor's medical guidance.

Tips on how to identify a seizure

Seizures can vary from momentary disruptions of the senses, to short periods of unconsciousness or staring spells, to uncontrollable muscle movements. Below is information about how to help identify the types of seizures you or your loved one can most likely experience—so you can talk to your doctor right away. The information provided is not intended to replace a doctor’s medical guidance.

Types of seizures common with TSC and how to identify them:

| INFANTILE SPASMS | FOCAL | GENERALIZED TONIC-CLONIC |
|---|--|--|
| <ul style="list-style-type: none">• Occur within first year of life, usually by 4-6 months• Typically seen as a sudden bending forward of the body with stiffening of the arms and legs lasting for a few seconds• Some infants arch their backs as they extend their arms and legs• Frequently occur in clusters upon awakening or going to sleep | <p>FOCAL AWARE</p> <ul style="list-style-type: none">• Used to be called simple partial seizures• Person is fully alert and able to interact• Experiences can include<ul style="list-style-type: none">—Involuntary motor movements on one side of the body—Intense sensory or emotional episodes, such as déjà vu or feeling unexplained emotions <p>FOCAL IMPAIRED AWARENESS</p> <ul style="list-style-type: none">• Used to be called complex partial seizures• Person loses awareness, may not respond, and has no memory of the seizure• Can vary between people, but may start with a blank stare, followed by chewing/lip smacking• Can also include random activity like picking at the air or clothes, attempting to pick up objects, remove clothing, repeat words or phrases, etc | <ul style="list-style-type: none">• Formerly known as “grand mal” seizures and is what most people think of when they hear the word seizure• Person loses consciousness or awareness, muscles extend and become rigid, and then muscles jerk rhythmically on both sides of the body |



Jen, at age 46
Living with TSC



| TONIC | ATONIC | MYOCLONIC | ATYPICAL ABSENCE |
|--|---|---|---|
| <ul style="list-style-type: none"> • Muscles in the body, arms, or legs suddenly become stiff or tense • May happen during sleep or can cause a person to fall if standing • Typically lasts for less than 20 seconds at a time | <ul style="list-style-type: none"> • May be referred to as a “drop attack” or “drop seizure” • Person has a sudden loss of muscle tone and goes limp • Mild seizures look like a head nod or drop, while severe seizures can cause a person to fall to the ground • People with these types of seizures may wear helmets to protect from injuries | <ul style="list-style-type: none"> • Sudden, brief shock-like muscle movements or jerks that usually don’t last more than a second or two • Person is alert • May occur in clusters and may be more pronounced upon waking | <ul style="list-style-type: none"> • Blank staring, with eye blinking, chewing movements, or lip smacking • Could include finger or hand rubbing, or other small hand movements • May begin and end gradually, usually lasting for about 5-30 seconds • May be difficult to distinguish between this seizure type and typical behavior in those with cognitive impairment |

You or your loved one may not experience all of these seizure types.

Seizures in childhood

Seizures usually start in childhood and can change over time

Although people with TSC can have their first seizure at any point during their lifetime, most people with TSC initially experience seizures in childhood.

THE MOST COMMON AGE
FOR SEIZURES TO START IS
AT APPROXIMATELY

3 MONTHS
OLD

NEARLY $\frac{2}{3}$ OF PEOPLE
HAVE THEIR FIRST SEIZURE

BEFORE
AGE 1

MORE THAN 80%
OF PEOPLE HAVE THEIR
FIRST SEIZURE

BEFORE
AGE 3

Seizures that happen during childhood may be subtle and may look very different from those that occur in adults. The first seizures experienced before the age of 2 are most commonly infantile spasms, focal seizures, or a combination of both.

Infantile spasms can be mistaken for colic, reflux, simple head nods, a startle reflex, or even normal body movements. At first onset, focal seizures can be very mild and increase in severity with time. *If you have a child with TSC and you suspect they have had a seizure, talk to your doctor immediately.*

Over time, infantile spasms may change to other seizure types. However, even if a child does not have infantile spasms, children may still experience other seizure types with age.

Seizures in adults with TSC

12% OF ADULTS

WITH TSC, AND NO HISTORY OF CHILDHOOD SEIZURES, CAN START HAVING SEIZURES IN ADULTHOOD

Adults with TSC can experience seizures even if they did not have one in childhood. Seizures may also stop and start again later in life or change into one of the other seizure types. Because people may not be diagnosed with TSC until adulthood if signs and symptoms are mild, it is important to be aware

of adult-onset seizures. It's important to write down any new symptoms of altered alertness, behavior, or motor movements to discuss with your doctor since these could be signs of potential new seizures.



In both children and adults:

Seizures can change in appearance, depending on where they originate in the brain and how they spread over time.

FOCAL AWARE SEIZURES

CAN
PROGRESS TO

FOCAL IMPAIRED AWARENESS

OR
POSSIBLY

GENERALIZED SEIZURES

It is important to monitor changes in your or your loved one's seizures so you can share them with your doctor. See the next page for support and resources to help track seizures.

The information provided is not intended to replace a doctor's medical guidance.





Maddie, at age 7
Living with TSC

Recognizing and tracking seizures

How to help your doctor identify the seizure type you or your loved one is experiencing

1. As difficult as it may seem in the moment, try to take a video to show your doctor if you suspect you or your loved one is having a seizure. This will help the doctor better identify the type of seizure and how to proceed with treatment.
2. Keep a diary of seizure activity in a dedicated notebook or by downloading a seizure tracking app. This can help you track to the best of your ability how often seizures happen and their possible triggers.

The information provided is not intended to replace a doctor's medical guidance.

Follow the seizure action plan you have created with your doctor to determine when to use rescue medication and when to visit the emergency department.

How your or your loved one's doctor may approach monitoring and treating seizures

There are many monitoring options and potential treatments used to reduce the frequency of seizures:



Electroencephalogram (EEG)

A test used to determine if there is abnormal brain activity that is causing seizures.



Magnetic resonance imaging (MRI)

An imaging technique used to look for noncancerous tumors that can occur with TSC that can lead to seizures.



Antiseizure medications

Therapies used to treat seizures.



Dietary approaches

Your or your loved one's doctor may prescribe a high-fat, low-carb medical diet that needs to be carefully monitored by a dietitian.



Vagus nerve stimulation

The use of a device that stimulates the vagus nerve.

Some people with TSC may not respond to these treatment approaches, and for some, surgery may be required to reduce seizure frequency. Ask your or your loved one's doctor for more information about these approaches.

It may be necessary to try more than one antiseizure medication at a time to effectively reduce the number of seizures because these drugs may work through different targets in the body. These therapies may take a while to start working, and it is not uncommon for people to be on multiple therapies at a time.



Where can I find support?

Finding out that you or a loved one has TSC can be overwhelming, but there is hope. While no two experiences are exactly alike, you are not alone.

There's a community of support and resources available to help you navigate life with TSC. If you or your loved one is newly diagnosed, we encourage you to connect with the TSC community of caregivers and experts who openly share their stories and advice.

Tuberous Sclerosis Complex Alliance

The Tuberous Sclerosis Complex Alliance (TSC Alliance) was founded in 1974 by four mothers in California who joined together to provide fellowship, generate awareness, pursue more knowledge, and provide hope to those who shared the common bond of TSC. These goals still drive the organization today. Its mission is to find a cure for TSC while improving the lives of those affected.

Learn more
about the
TSC community
by visiting

TSCalliance.org



@tscalliance

Jazz Pharmaceuticals

At Jazz Pharmaceuticals, we are dedicated to offering innovative solutions that go beyond medicine. Our mission is to transform the lives of children, adults, and families who live with rare neurological diseases. Families are at the center of everything we do.