



Tuberous sclerosis complex





This guide is not meant to take the place of medical advice.

Please consult with your doctor about your genetic results and health care choices. The information in this guide was up to date at the time it was written in 2019. But new information may come to light with new research. You may find it helpful to share this guide with friends and family members or doctors and teachers of the person who has tuberous sclerosis complex.



What is tuberous sclerosis complex?



Tuberous sclerosis complex happens when there are changes to the TSC1 gene or the TSC2 gene. These changes can keep these genes from working as they should.

Key role

The TSC1 and TSC2 genes play a key role in controlling cell growth.

Symptoms

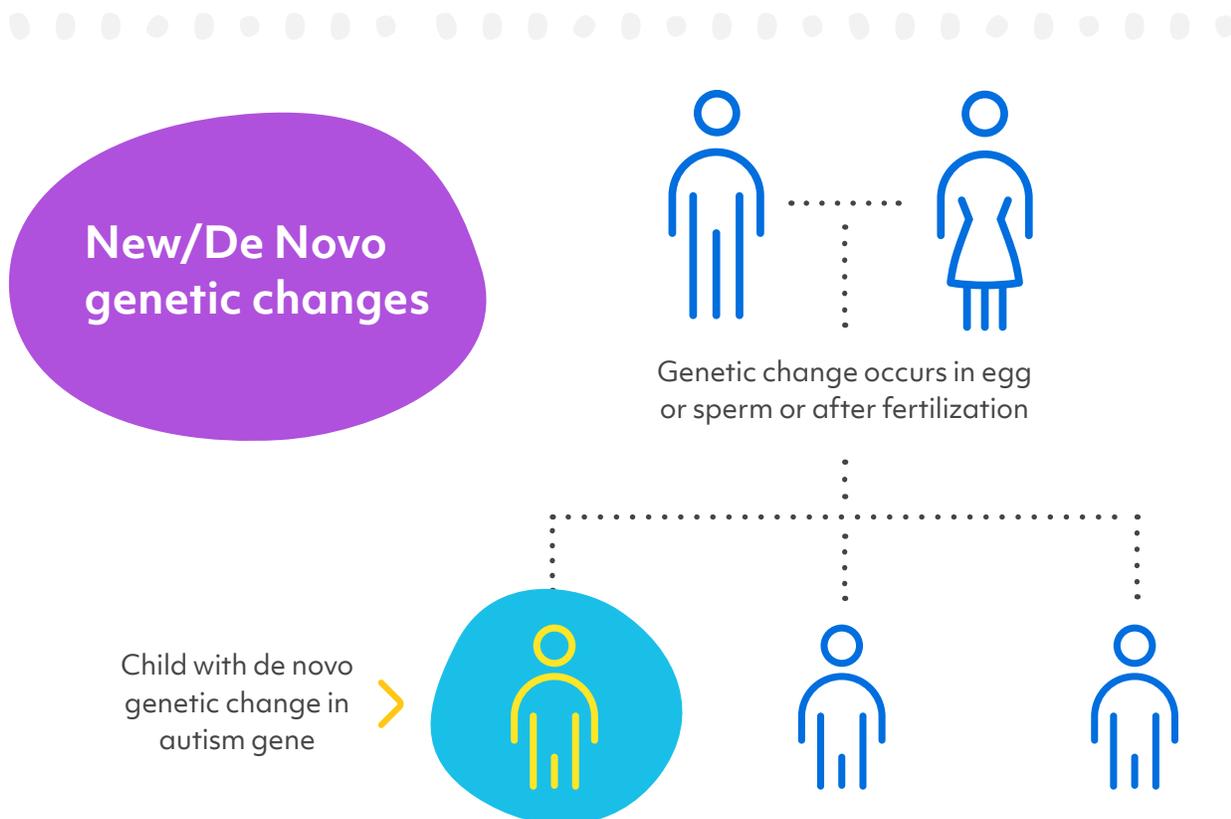
Tuberous sclerosis complex usually affects the central nervous system. It results in several symptoms. Changes in the TSC1 and TSC2 genes can lead to the growth of noncancerous tumors in different organs. Many people who have tuberous sclerosis complex have:

- Seizures
- Developmental delay
- Intellectual disability
- Autism, or behavior problems, or both
- Changes in the appearance of skin

What causes tuberous sclerosis complex?

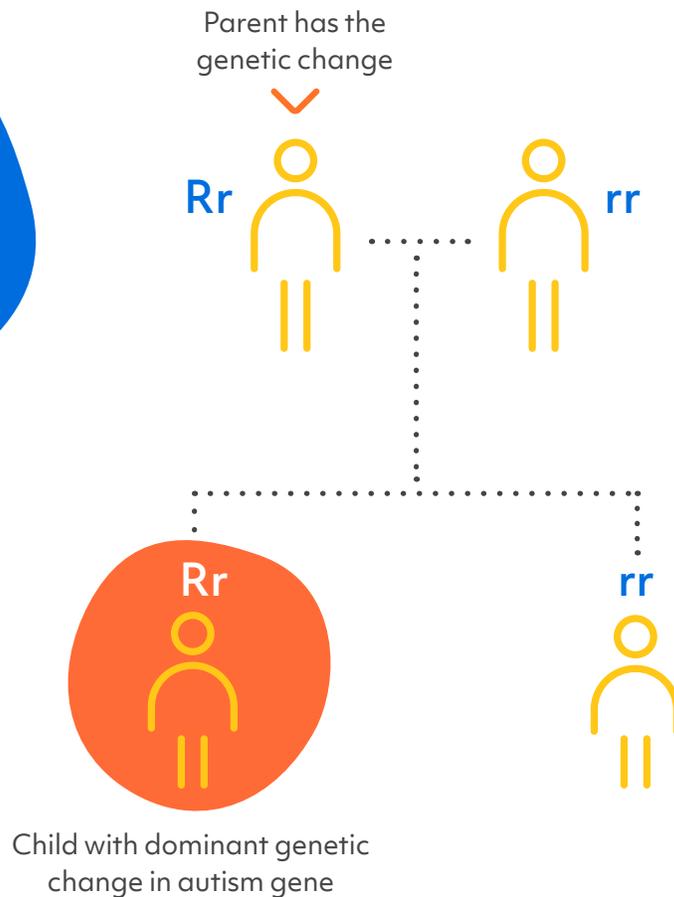
Our genes contain the instructions, or code, that tell our cells how to grow, develop, and work. Every child gets two copies of the TSC1 and TSC2 genes: one copy from their mother, from the egg, and one copy from their father, from the sperm. In most cases, parents pass on exact copies of the gene to their child. But the process of copying genes is not perfect. A change in the genetic code can lead to physical issues, developmental issues, or both.

Sometimes a random change happens in the sperm or egg. This change to the genetic code is called a 'de novo', or new, change. The child can be the first in the family to have the gene change.



Dominant inheritance

Children have a 50% chance of inheriting the genetic change



De novo changes can take place in any gene. We all have some de novo changes, most of which don't affect our health. But because the TSC1 and TSC2 genes play a key role in development, de novo changes in these genes can have a meaningful effect.

Research shows that tuberous sclerosis complex is often the result of a de novo change in the TSC1 or TSC2 genes. Many parents who have had their genes tested do not have the gene change found in their child who has the condition. In some cases, tuberous sclerosis complex happens because the gene change was passed down from a parent.

Why does my child or I have a change in the TSC1 or TSC2 genes?

No parent causes their child's tuberous sclerosis complex. We know this because no parent has any control over the gene changes that they do or do not pass on to their children. Please keep in mind that nothing a parent does before or during the pregnancy causes this to happen. The gene change takes place on its own and cannot be predicted or stopped.





What are the chances that other family members or future children will have tuberous sclerosis complex?

Each family is different. A geneticist or genetic counselor can give you advice on the chance that this will happen again in your family.

The risk of having another child who has tuberous sclerosis complex depends on the genes of both birth parents.

- If neither birth parent has the same gene change found in their child, the chance of having another child who has the syndrome is on average 1 percent. This 1 percent chance is higher than the chance of the general population. The increase in risk is due to the very unlikely chance that more of the mother's egg cells or the father's sperm cells carry the same change in the gene.
- If one birth parent has the same gene change found in their child, the chance of having another child who has the syndrome is 50 percent.

For a symptom-free sibling, a brother or sister, of someone who has tuberous sclerosis complex, the risk of having a child who has the condition depends on the symptom-free sibling's genes and their parents' genes.

- If neither parent has the same gene change found in their child who has the condition, the symptom-free sibling has a nearly 0 percent chance of having a child who has tuberous sclerosis complex.
- If one birth parent has the same gene change found in their child who has tuberous sclerosis complex, the symptom-free sibling has a small chance of also having the same gene change. If the symptom-free sibling has the same gene change as their sibling who has the syndrome, the symptom-free sibling's chance of having a child who has tuberous sclerosis complex is 50 percent.

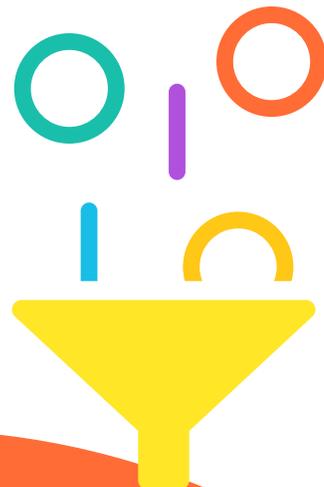
For a person who has tuberous sclerosis complex, the risk of having a child who has tuberous sclerosis complex is about 50 percent.

How many people have tuberous sclerosis complex?

An estimated 40,000 Americans and at least 2 million people in the world have tuberous sclerosis complex. This is about 1 in 6,000 to 1 in 9,000 people.

Do people who have tuberous sclerosis complex look different?

Most people who have tuberous sclerosis complex do not look very different. More than 90 percent of people who have the condition have small light spots on their skin, known as ash leaf spots. Some people have small bumps on their face around the nose or mouth, called angiofibromas.



How is tuberous sclerosis complex treated?

The growths, or tumors, that result from tuberous sclerosis complex are not cancerous. But they can still cause serious problems.

There are many options to treat the growths that are associated with tuberous sclerosis complex. If doctors think a growth is causing seizures, they may perform surgery. In other cases, medication may be used to treat the seizures.

Research shows that controlling seizures as early as possible can improve learning. If seizures happen, consult a neurologist. There are many types of seizures, and not all types are easy to spot. To learn more, you can refer to resources such as the Epilepsy Foundation's website: www.epilepsy.com/learn/types-seizures.

A genetic diagnosis can help people decide on the best way to track the condition and manage therapies. Doctors can refer people to specialists for:

- Physical exams and brain studies
- Genetics consults
- Development and behavior studies
- Study and treatment of seizures
- Kidney checks. There is a small risk of kidney cysts and kidney cancer. A kidney doctor is needed to check kidney function.
- Other issues, as needed

A developmental pediatrician, neurologist, or psychologist can follow progress over time and can help:

- Suggest the right therapies. This can include physical, occupational, speech, or behavioral therapy.
- Guide individualized education plans (IEPs).
- Treat attention disorders.

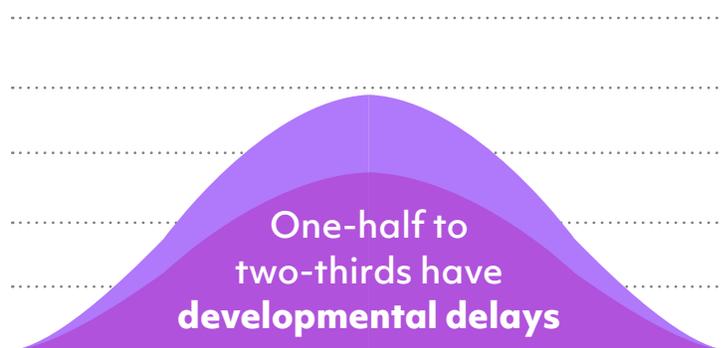
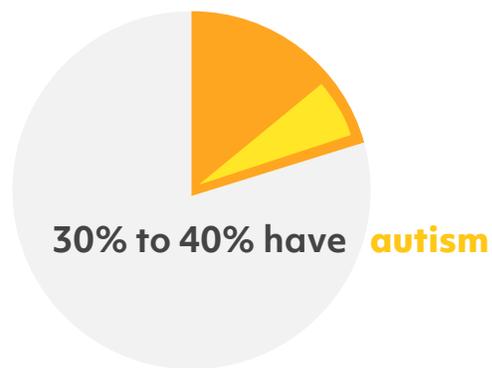
Specialists advise that therapies for tuberous sclerosis complex should begin as early as possible, ideally before a child begins school.

This section includes a summary of information from major published studies. To learn more about the articles, see the [Sources and references](#) section of this guide.

Behavior and development concerns linked to tuberous sclerosis complex

Behavior

About 30 percent to 40 percent of people who have the condition have **autism**. A similar percentage have attention deficit hyperactivity disorder, also called **ADHD**. Many people show disruptive behaviors including aggression, anxiety, and self-injury.



Learning

About one-half to two-thirds of people who have the condition have **developmental delays**. These range from mild learning disabilities to severe intellectual disability.

Medical and physical concerns linked to tuberous sclerosis complex

The **tumors** that result from tuberous sclerosis complex are not usually cancer. But they can still cause serious problems.



Brain

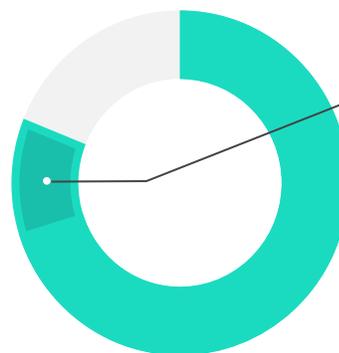
People who have tuberous sclerosis complex may have brain growths called tubers. Cortical tubers generally form on the surface of the brain, but they may also appear in deep areas of the brain. If a growth causes seizures, surgery may be needed to permanently remove the growth. Most people who have the condition will experience epilepsy at some point in their life.



Kidney

70 percent to 80 percent of people who have tuberous sclerosis complex have cysts and non-cancerous tumors in their kidneys. Cysts are usually small and few. About 20 percent of people who have tuberous sclerosis complex develop lots of these cysts, which can lead to more serious kidney problems.

Benign kidney tumors do not usually produce symptoms. But they can sometimes grow so large that they cause pain and, eventually, kidney failure. Rarely — less than 5 percent of the time — one of these growths becomes cancerous.



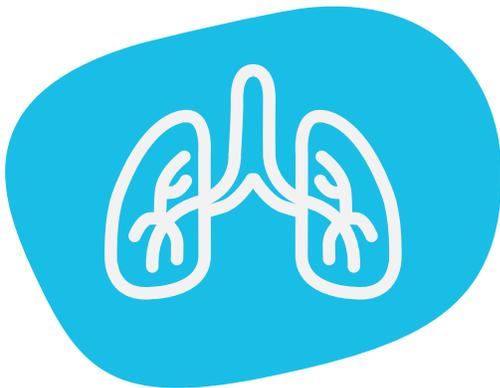
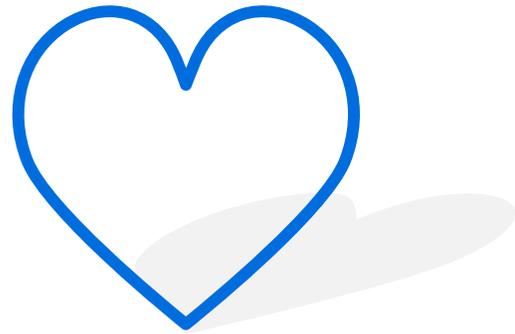
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Medical and physical concerns linked to tuberous sclerosis complex

Heart

Tumors in the heart are usually largest at birth and get smaller as the person gets older. These heart tumors are called cardiac rhabdomyomas. If they block the flow of blood at birth or lead to severe changes in heart rhythm, they can cause problems.



Lungs

Growth of smooth muscle cells in the lungs can lead to cysts and shortness of breath. These growths are known as lymphangioleiomyomatosis. They happen most often in adult women.

Eyes

Although growths in the eyes are uncommon, they may occur. They can cause problems if they grow and block too much of the retina.





**Where can I
find support
and resources?**

Tuberous Sclerosis Complex International
www.tscinternational.org



Sources and References

The content in this guide comes from published studies about tuberous sclerosis complex. Below you can find details about each study, as well as links to summaries or, in some cases, the full article.

- National Institute of Neurological Disorders and Stroke, Tuberous Sclerosis Fact Sheet
www.ninds.nih.gov/Disorders/Patient-Caregiver-Education/Fact-Sheets/Tuberous-Sclerosis-Fact-Sheet#3220_1
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- Northrup H. *et al. GeneReviews*, (2018). Tuberous Sclerosis Complex
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