CONDITION

Tuberous sclerosis complex

Tuberous sclerosis complex is a genetic condition that causes noncancerous growths in the brain, skin, kidneys, lungs, heart, and other organs.

WHAT IT IS
Genetic condition causing noncancerous growths in multiple organs

BODY SYSTEM
Brain, skin, kidneys, lungs, heart, and eyes

MAIN SYMPTOMS
Seizures, skin changes, learning differences, kidney problems

WHAT CAUSES IT
Inherited or new changes in TSC1 or TSC2

HOW IT IS DIAGNOSED
Clinical criteria or TSC1 and TSC2 genetic testing

MAIN TREATMENT
Lifelong monitoring, symptom treatment, and mTOR-inhibitor medicines

CAN IT BE CURED?
No - the genetic change remains lifelong

IS IT AN EMERGENCY?
Sometimes - prolonged seizures or sudden breathing difficulty

LONG-TERM OUTLOOK
Many live active lives with lifelong monitoring

Overview

Tuberous sclerosis complex, or TSC, is a lifelong genetic condition. It can cause noncancerous growths in several parts of the body, especially the brain, skin, kidneys, heart, lungs, and eyes.

These growths are often called hamartomas. A hamartoma is an overgrowth of cells that normally belong in that part of the body. Most are not cancer, but they can still cause problems if they grow, bleed, or press on nearby tissue.

TSC affects each person differently. Some people have mild skin changes and few other concerns. Others have seizures, kidney disease, lung problems, or significant learning and developmental needs. People in the same family can be affected in different ways.

The name comes from changes sometimes seen in the brain. "Tuberous" refers to growths

called tubers, while "sclerosis" means hardened tissue. TSC is not contagious and is not caused by anything someone did during pregnancy.

Regular monitoring matters because some complications can develop before they cause clear symptoms. Finding them early may allow treatment before lasting harm occurs.

Symptoms

Signs can appear before birth, during childhood, or in adulthood. Some growths are found by chance during a scan for another reason.

Common features include:

Not everyone has all these features. Normal intelligence is possible, and some people do not discover they have TSC until adulthood.

- Seizures: These may begin in infancy or later. Infantile spasms are brief, repeated movements that often happen in clusters.
- Developmental or behavioural differences: These can include learning disability, autism, attention problems, anxiety, low mood, sleep problems, or difficulty managing emotions.
- Skin changes: Pale patches, small reddish facial bumps, thickened areas, or growths around the nails may develop.
- Kidney changes: Fluid-filled cysts and noncancerous growths called angiomyolipomas can occur. These may cause pain, bleeding, high blood pressure, or reduced kidney function.
- Brain growths: Some people develop subependymal giant cell astrocytomas, or SEGAs. These noncancerous growths can block the normal flow of fluid around the brain.
- Heart growths: Cardiac rhabdomyomas often form before birth or in infancy. Many shrink over time, but some affect heart rhythm or blood flow.
- Lung changes: Lymphangioleiomyomatosis, or LAM, mainly affects adult women with TSC. It can cause shortness of breath, collapsed lung, cough, or chest discomfort.
- Eye and mouth changes: Small retinal growths, pits in tooth enamel, and gum growths may occur.

Causes and risk factors

TSC is caused by a harmful change in either the TSC1 or TSC2 gene. These genes normally help control cell growth through a cell-signalling system called the mTOR pathway. When one

gene does not work properly, cells can grow when they should not.

TSC follows an autosomal dominant inheritance pattern. This means a person with TSC has a 1 in 2 chance of passing the altered gene to each child. The chance is the same for every pregnancy.

Many people with TSC are the first in their family to have it. In these cases, the genetic change arose in an egg, sperm, or early embryo rather than being inherited from a parent.

Some people have mosaic TSC. This means the gene change is present in only some of their cells. Mosaicism may cause milder or less typical features and can make genetic testing more complex.

The altered gene does not reliably predict how strongly someone will be affected. Even close relatives with the same genetic change may have different symptoms.

Diagnosis

TSC may be diagnosed from a combination of characteristic features. International diagnostic criteria group these as major and minor features. A definite clinical diagnosis generally requires either two major features or one major feature with at least two minor features.

A disease-causing change in TSC1 or TSC2 can also confirm the diagnosis. However, a negative genetic test does not completely rule out TSC. Current testing may miss some changes, particularly mosaic ones.

Assessment may include:

The tests needed depend on age, sex, symptoms, and previous findings. Genetic counselling can help explain test results and what they may mean for relatives.

- A skin examination, sometimes using ultraviolet light to reveal pale patches
- MRI scans of the brain and abdomen
- Kidney function tests, urine tests, and blood pressure checks
- An electroencephalogram, or EEG, which records electrical activity in the brain
- Heart rhythm testing and an ultrasound scan of the heart
- Eye, dental, developmental, and mental health assessments
- Lung testing and chest imaging when appropriate

Treatment and management

There is no treatment that removes the underlying genetic change. Care focuses on monitoring affected organs, controlling symptoms, and treating growths that could cause harm.

A coordinated team may include specialists in neurology, kidneys, lungs, heart care, skin, genetics, development, mental health, and surgery.

Seizures are treated with antiseizure medicines. Infantile spasms need rapid specialist assessment because early control may support development. If seizures remain difficult to control, options may include epilepsy surgery, dietary treatment, nerve stimulation, or an mTOR-inhibitor medicine.

mTOR inhibitors, such as everolimus or sirolimus, block the overactive cell-growth pathway involved in TSC. Depending on the problem and local approval, they may be used for growing SEGAs, kidney angiomyolipomas, certain difficult-to-control seizures, LAM, or facial skin growths. These medicines require monitoring because they can cause side effects such as mouth ulcers, infection, or changes in blood test results.

A SEGA may need treatment if it is growing or blocking brain fluid. Treatment can involve an mTOR inhibitor or surgery.

Kidney care aims to preserve as much healthy tissue as possible. Treatment for a growing or bleeding angiomyolipoma may include an mTOR inhibitor, blocking its blood supply through embolisation, or kidney-sparing surgery. Blood pressure and kidney function also need ongoing review.

Heart rhabdomyomas often shrink without treatment. Intervention is considered when they interfere with blood flow, heart function, or rhythm.

Skin growths may be managed with topical mTOR treatment, laser therapy, or surgery. Developmental therapies, educational support, behavioural care, sleep treatment, and mental health care are also important parts of TSC management.

Complications

Complications depend on which organs are affected.

Seizures can disrupt learning, safety, and daily life. A prolonged seizure can become status epilepticus, which is a medical emergency. TSC-associated neuropsychiatric disorders, often shortened to TAND, can affect education, relationships, employment, and independence.

A growing SEGA can block the flow of fluid in the brain. This can raise pressure inside the skull and cause worsening headaches, vomiting, vision changes, unusual sleepiness, or changes in

behaviour.

Kidney angiomyolipomas may bleed, sometimes suddenly. Kidney cysts, high blood pressure, and repeated procedures can also reduce kidney function over time.

LAM can gradually affect breathing. Lung cysts may rupture and cause a collapsed lung, known as a pneumothorax. This usually causes sudden chest pain or shortness of breath.

Heart growths can occasionally obstruct blood flow or cause an abnormal rhythm, particularly early in life. Most become smaller with age.

Prevention

TSC itself cannot currently be prevented. It results from a genetic change that is present from conception or develops very early after conception.

If you have TSC or a family history of it, genetic counselling can explain inheritance and reproductive options. These may include testing during pregnancy or testing embryos created through in vitro fertilisation. These are personal choices, and counselling should be non-directive.

Complications cannot always be prevented, but regular surveillance can identify changes before they cause major symptoms. Keeping appointments for brain, kidney, lung, heart, developmental, eye, skin, and dental checks is an important part of care.

Following a seizure plan, taking prescribed medicines consistently, and avoiding smoking can also reduce specific risks. These steps manage TSC and its complications; they do not prevent the genetic condition itself.

Living with it and what to expect

TSC is lifelong, but its effects can change with age. Heart rhabdomyomas often shrink during childhood. Seizures and developmental concerns commonly appear early, while kidney growths and lung disease may become more important during adulthood. Skin findings may become more noticeable over time.

Regular monitoring usually continues even when you feel well. Scans and other checks are intended to find silent changes, such as a growing kidney angiomyolipoma or brain growth. The schedule is based on age, existing findings, symptoms, and previous treatment.

Many people with TSC attend school, work, have relationships, and raise families. Some need limited adjustments, while others need lifelong help with communication, personal care,

decision-making, or seizure safety.

Mental health and behavioural symptoms deserve the same attention as physical problems. Anxiety, low mood, sleep difficulty, attention problems, and family stress are not simply things you must accept as part of TSC.

The long-term outlook is influenced by seizure control, learning needs, kidney health, lung involvement, and access to monitoring and treatment. New growths or symptoms can appear, but this does not mean everyone's condition steadily worsens.

When to get medical help

Call emergency services if someone with TSC has:

A first seizure also needs prompt medical assessment.

Arrange a routine or prompt follow-up if you notice new seizures, changes in seizure pattern, worsening headaches, loss of skills, major changes in behaviour, new breathing limits, persistent kidney-area pain, blood in the urine, or a changing skin growth.

Routine TSC appointments remain important even when there are no new symptoms. Contact the care team if you are unsure when the next brain, kidney, lung, heart, or developmental review is due.

- A seizure lasting five minutes or longer, unless their seizure plan gives an earlier threshold
- Repeated seizures without recovering between them
- Sudden severe shortness of breath or chest pain
- Severe abdominal or side pain with faintness, weakness, or visible blood in the urine
- A severe or rapidly worsening headache with vomiting, unusual sleepiness, confusion, or vision changes

Common questions

Is tuberous sclerosis complex cancer?

No. Most TSC growths are noncancerous. They can still need treatment if they grow, bleed, block fluid, or affect how an organ works. Any unusual or changing mass should be assessed rather than assumed to be part of TSC.

Can tuberous sclerosis complex be mild?

Yes. Some people have only a few skin findings or growths that never cause major problems.

Others have more extensive needs. Mild symptoms do not remove the need for monitoring because internal changes may not be noticeable.

Will my child inherit TSC from me?

If you have a disease-causing TSC1 or TSC2 change, each child has a 1 in 2 chance of inheriting it. A child who inherits the change may be affected more mildly or more strongly than their parent.

Does TSC cause autism or learning disability?

TSC can be associated with autism, intellectual disability, attention problems, language delay, anxiety, and other behavioural or emotional concerns. It does not affect everyone in the same way, and some people have typical learning and development.

Can someone with TSC have children?

Yes. Pregnancy planning may include genetic counselling, a medicine review, and assessment of kidney, heart, seizure, and lung health. Some TSC medicines may need specialist review before conception or during pregnancy.

Does TSC get worse with age?

Not always. Some features improve, while others emerge or grow later. Heart growths often shrink, but kidney and lung changes may become more relevant in adulthood. Lifelong monitoring helps identify these changes early.

Sources

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