CONDITION

Tuberous sclerosis

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

WHAT IT IS
Genetic condition causing noncancerous growths in multiple organs

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

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

WHAT CAUSES IT
Inherited or new changes in TSC1 or TSC2

HOW IT IS DIAGNOSED
Clinical criteria and genetic testing

MAIN TREATMENT
Symptom treatment, mTOR medicines, surgery, and lifelong monitoring

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

IS IT AN EMERGENCY?
Sometimes, with prolonged seizures or severe breathing difficulty

LONG-TERM OUTLOOK
Many live active lives with tailored lifelong care

Overview

Tuberous sclerosis, also called tuberous sclerosis complex or TSC, is a lifelong genetic condition. "Tuberose sclerosis" is another spelling you may see, although tuberous sclerosis is the standard medical name.

TSC can cause growths to form in different organs. These growths are usually noncancerous, meaning they do not spread through the body like cancer. However, their size and location can still cause health problems.

The brain, skin, kidneys, heart, lungs, and eyes are most often affected. Some people have only a few mild features. Others have seizures, learning or behavioural differences, kidney problems, or several affected organs.

Features may appear at different ages. Heart growths can sometimes be found before birth, while kidney or lung changes may not develop until adulthood. This is why people with TSC usually need regular checks even when they feel well.

Symptoms

TSC does not look the same in everyone, including people in the same family. Symptoms depend on which organs are involved.

Brain changes may cause:

Doctors may group behavioural, learning, mental health, and thinking differences under the term TSC-associated neuropsychiatric disorders , or TAND. These concerns can be overlooked, so regular screening is important.

Skin findings are common. They may include pale patches, small reddish bumps on the face, thickened areas of skin, or growths around the fingernails and toenails. Pale patches can be easier to see under a special ultraviolet light. Skin changes are not usually dangerous, but they may bleed, catch on clothing, or affect confidence.

Kidney changes include fluid-filled cysts and growths called angiomyolipomas . Angiomyolipomas contain blood vessels, muscle, and fat. They often cause no symptoms, but larger growths can bleed or affect kidney function.

Some people develop a lung condition called lymphangioleiomyomatosis , or LAM. It mainly affects women after puberty and can cause breathlessness, cough, chest pain, or a collapsed lung.

Heart growths called rhabdomyomas are most often found in babies and children. They frequently become smaller with age. Occasionally, they affect heart rhythm or blood flow.

Eye growths usually do not disturb vision, although eye examinations are still part of care.

- Seizures, including infantile spasms in babies
- Learning difficulties or intellectual disability
- Autism, attention difficulties, anxiety, sleep problems, or mood changes
- Headaches, vomiting, or balance problems if a brain growth blocks fluid flow

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 this control is disrupted, cells can grow into the characteristic TSC growths.

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 the egg, sperm, or early embryo rather than being inherited from a parent. It is not caused by anything a parent did during pregnancy.

Having the same genetic change does not mean relatives will have the same symptoms. The organs affected and the severity of TSC can differ greatly.

Diagnosis

TSC may be suspected because of seizures, characteristic skin findings, kidney growths, a heart rhabdomyoma, or findings on a pregnancy scan. Sometimes it is identified after a relative receives a diagnosis.

Diagnosis is based on internationally agreed clinical criteria. These criteria combine major and minor features seen on examinations and imaging. A definite diagnosis can be made from a specific combination of these features.

Genetic testing can also confirm TSC if it finds a disease-causing change in TSC1 or TSC2. A negative genetic test does not always rule it out because current testing cannot detect every possible genetic change.

Tests used to assess the condition may include:

The assessment also includes development, learning, behaviour, mental health, and daily functioning. Testing is tailored to age, symptoms, and previous findings.

- MRI scans of the brain and abdomen
- Skin, dental, and eye examinations
- An electroencephalogram, or EEG, to measure electrical activity in the brain
- Kidney function tests and blood pressure checks
- Heart rhythm tests and an ultrasound scan of the heart
- Lung scans and breathing tests when appropriate

Treatment and management

There is no treatment that removes the underlying genetic change. Care aims to control symptoms, protect organ function, support development, and identify problems early.

Seizures are treated with anti-seizure medicines. Infantile spasms are a particular seizure type in babies and need prompt specialist assessment; vigabatrin is commonly used as initial treatment when the spasms are linked to TSC. Other medicines, dietary therapy, implanted devices, or epilepsy surgery may be considered when seizures remain difficult to control.

Medicines called mTOR inhibitors , including everolimus or sirolimus, act on the overactive cell-growth pathway involved in TSC. Depending on the organ involved, they may be used to shrink certain brain or kidney growths, help control LAM, or treat some skin growths. These medicines require medical supervision and blood tests because they can cause effects such as mouth ulcers, infections, or changes in blood fats.

A growing brain lesion called a subependymal giant cell astrocytoma , or SEGA, may be treated with an mTOR inhibitor or surgery. Urgent surgery may be needed if it blocks the normal flow of fluid around the brain.

Kidney angiomyolipomas may be monitored, treated with an mTOR inhibitor, or managed with a procedure that blocks their blood supply. Kidney-sparing approaches are preferred when possible. Active bleeding requires urgent treatment.

Skin growths can sometimes be managed with topical mTOR treatment, laser therapy, or surgery. Treatment may help with bleeding, irritation, or appearance.

Support can also include speech and language therapy, occupational therapy, educational adjustments, psychological care, and help with sleep or behaviour. TAND concerns should be reviewed regularly rather than only when a crisis occurs.

Complications

Seizures can affect safety, learning, sleep, and independence. Frequent or prolonged seizures may be harder to control, but early recognition and specialist treatment can improve management.

A SEGA can block fluid drainage in the brain, causing a dangerous rise in pressure. Warning signs can include a new or worsening headache, repeated vomiting, unusual sleepiness, vision changes, or loss of balance.

Kidney angiomyolipomas can bleed. TSC can also lead to high blood pressure or reduced kidney function over time. Kidney cancer is uncommon, but unusual or changing kidney

masses need careful assessment.

LAM can gradually reduce lung function. A lung cyst may rupture and cause a collapsed lung, leading to sudden chest pain and breathlessness.

Mental health, learning, sensory, and behavioural concerns may affect education, work, relationships, and quality of life. These are part of TSC and deserve the same attention as physical symptoms.

Prevention

TSC cannot usually be prevented because it is caused by a genetic change. Lifestyle choices do not cause it.

If you have TSC or a known family gene change, genetic counselling can explain inheritance and testing options. These may include testing during pregnancy or testing embryos created through in vitro fertilisation. These are personal choices, and counselling should provide information without directing your decision.

Regular monitoring does not prevent TSC itself, but it can reduce the effect of some complications by finding changes before they cause major symptoms. Keeping scheduled brain, kidney, lung, eye, skin, dental, heart, and developmental reviews is an important part of care.

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 differences commonly appear early, while kidney growths may enlarge over years. Lung involvement usually appears later and mainly affects women.

Some people need little day-to-day treatment beyond routine monitoring. Others need several medicines, procedures, learning support, or help with daily activities. New concerns can appear even after a stable period, so follow-up continues through adulthood.

Most care is planned rather than urgent. The schedule is based on age, sex, organ involvement, symptoms, and previous test results. Moving from children's to adult services should include a clear record of imaging, genetic results, seizure history, medicines, and support needs.

School or workplace adjustments may help with seizures, fatigue, attention, sensory needs, or learning. A written seizure plan can help family members, schools, or colleagues know what to

do.

The long-term outlook is broad. Many people study, work, have relationships, and live independently. Others need lifelong practical support, particularly when epilepsy or intellectual disability is severe. Regular specialist care aims to protect health and support as much independence as possible.

When to get medical help

Arrange routine follow-up

Contact your usual care team if there are new or gradually changing skin growths, headaches, seizures, behaviour, learning, breathing, urinary, or vision concerns. Also seek review if medicines are causing side effects or scheduled surveillance has been missed.

Seek prompt medical advice

Ask for prompt assessment after a first suspected seizure, a clear change in seizure pattern, blood in the urine, persistent flank pain, increasing breathlessness, or repeated headaches and vomiting. Babies with clusters of brief body stiffening, head drops, or sudden repeated jerks need urgent assessment for infantile spasms.

Get emergency help

Call emergency services for:

Follow an existing emergency seizure plan if one has been provided.

- A seizure lasting five minutes or longer
- Repeated seizures without recovery between them
- Severe breathing difficulty or sudden chest pain with breathlessness
- Sudden severe side or back pain with faintness or weakness
- New severe headache with repeated vomiting, confusion, marked sleepiness, or collapse

Common questions

Is tuberous sclerosis cancer?

No. Most TSC growths are noncancerous. They can still cause problems by bleeding, damaging organ function, or pressing on nearby structures. Cancer can occur, but it is not the usual nature of TSC growths.

Can tuberous sclerosis be mild?

Yes. Some people have few symptoms and are diagnosed only after a scan, skin examination, or relative's diagnosis. Mild symptoms do not remove the need for monitoring because kidney, brain, or lung changes can develop without early warning signs.

Will my child inherit tuberous sclerosis?

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 differently from you. Genetic counselling can explain testing and reproductive options.

Does tuberous sclerosis get worse with age?

Not in one predictable way. Some findings, such as heart rhabdomyomas, often shrink. Kidney growths may enlarge, and lung changes can appear in adulthood. Seizures, learning needs, and behaviour may also change over time.

Can someone with TSC live independently?

Many people can, especially when learning and physical effects are mild. Others need support with education, employment, health care, money, or daily activities. Independence is shaped by seizure control, development, mental health, and organ complications.

Is tuberose sclerosis the same condition?

"Tuberose sclerosis" is sometimes used as an alternative spelling. The standard medical terms are tuberous sclerosis and tuberous sclerosis complex , usually shortened to TSC.

Sources

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