CONDITION

Tuberous sclerosis-1

Tuberous sclerosis-1 is an inherited condition that can cause noncancerous growths in several organs.

WHAT IT IS
Inherited condition causing noncancerous growths in several 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 harmful change in TSC1

HOW IT IS DIAGNOSED
Clinical criteria, imaging, and TSC1 genetic testing

MAIN TREATMENT
Monitoring, symptom treatment, targeted medicines, and procedures

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

IS IT AN EMERGENCY?
Sometimes - seizure lasting five minutes or more

LONG-TERM OUTLOOK
Many lead active lives with lifelong specialist monitoring

Overview

Tuberous sclerosis-1 is a form of tuberous sclerosis complex caused by a harmful change in a gene called TSC1 . This gene helps control how cells grow. When it does not work as expected, cells can grow into noncancerous masses in different parts of the body.

These growths are often called hamartomas . They are not cancer, but their size or location can cause problems. The brain, skin, kidneys, heart, lungs, and eyes may be affected. The number and severity of problems differ greatly, even among relatives with the same genetic change.

Tuberous sclerosis-1 is lifelong. Some features appear before birth or during infancy, while others develop in childhood or adulthood. Regular monitoring matters because kidney, brain, or lung changes may grow without causing early symptoms.

TSC1-related tuberous sclerosis is sometimes less severe on average than the form caused by changes in the TSC2 gene. This does not predict what will happen to one person. Either form can cause significant health and developmental needs.

Symptoms

The signs of tuberous sclerosis-1 depend on which organs are affected. Some people have only a few visible features. Others need ongoing care for seizures, development, kidney growths, or other complications.

Changes involving the brain can include seizures, learning difficulties, developmental delay, or changes in attention, mood, sleep, and behaviour. Seizures may begin during infancy, sometimes as brief clusters of head drops or body stiffening called infantile spasms .

Brain imaging may show cortical tubers, which are areas where brain tissue developed differently. It may also show nodules along the fluid-filled spaces of the brain. A small number of these nodules grow into a subependymal giant cell astrocytoma , or SEGA. This is a noncancerous brain tumour, but it can block the flow of brain fluid and raise pressure inside the skull.

Common skin features include:

Kidney changes can include cysts and angiomyolipomas , which are growths made of blood vessels, muscle, and fat. They may cause no symptoms, but larger growths can bleed or affect kidney function.

Heart growths called rhabdomyomas may be found before birth or during infancy. Many shrink with age. Occasionally, they interfere with blood flow or the heart's rhythm.

Lung involvement, especially lymphangioleiomyomatosis or LAM , occurs mainly in women after puberty. It can cause shortness of breath, cough, chest pain, or a collapsed lung. Eye growths are also possible, although they often do not affect vision.

- Pale patches that may be easier to see under a special ultraviolet light
- Small reddish bumps on the face, called facial angiofibromas
- Thickened, rough skin, often on the lower back
- Firm growths around or beneath the nails

Causes and risk factors

Tuberous sclerosis-1 is caused by a harmful change, also called a pathogenic variant, in the TSC1 gene. This gene provides instructions for making hamartin, a protein that helps prevent cells from growing and dividing when they should not.

A TSC1 change allows a cell-growth pathway called mTOR to become overactive. This contributes to the growths seen in tuberous sclerosis.

The condition follows an autosomal dominant inheritance pattern. This means one altered copy of TSC1 is enough to cause it. A person with the condition has a 1 in 2 chance of passing the altered gene to each child. The child's features cannot be predicted from the parent's symptoms.

Many people are the first in their family to have the condition. In these cases, the genetic change arose in an egg, sperm, or early embryo. It was not caused by anything a parent did during pregnancy.

Occasionally, a parent has the TSC1 change in only some of their cells. This is called mosaicism. It can make genetic testing and estimates of recurrence in a family more complex.

Diagnosis

Tuberous sclerosis can be diagnosed using established clinical criteria. These criteria combine characteristic findings from skin examinations, eye examinations, scans, and other tests. A definite clinical diagnosis does not require every organ to be affected.

Testing may include:

Genetic testing can confirm the TSC1 subtype by finding a disease-causing TSC1 variant. It may also help relatives understand whether they need testing. If testing finds a TSC2 variant, the condition is not classified as tuberous sclerosis-1.

A negative genetic test does not always rule out tuberous sclerosis. Current tests cannot identify every possible genetic change, and a clinical diagnosis may still be made when the characteristic findings are present.

- MRI of the brain to look for cortical tubers, nodules, or a SEGA
- Kidney MRI or other imaging to check for angiomyolipomas and cysts
- Skin, dental, and eye examinations
- An electroencephalogram, or EEG, to measure electrical activity in the brain
- Heart imaging and an electrocardiogram to assess structure and rhythm
- Lung tests and chest imaging when appropriate

Treatment and management

There is no treatment that removes the TSC1 genetic change. Care focuses on controlling symptoms, treating growths that are causing harm, and finding complications early.

Monitoring usually continues throughout life. The timing of brain, kidney, heart, eye, skin, and lung checks is based on age, previous findings, and symptoms. Care may involve neurology, kidney, heart, lung, skin, genetics, mental health, and developmental specialists.

Seizures are treated with antiseizure medicines. Infantile spasms need prompt specialist assessment because early control can influence development. For seizures that remain difficult to control, options may include additional medicines, epilepsy surgery, dietary therapy, nerve stimulation, or an mTOR-blocking medicine.

Medicines called mTOR inhibitors reduce activity in the overactive cell-growth pathway. They may be used for certain growing brain tumours, kidney angiomyolipomas, seizures, lung involvement, or skin lesions. Benefits and side effects require regular review and blood tests.

A SEGA may be treated with surgery or an mTOR inhibitor when it is growing, causing symptoms, or blocking brain-fluid flow. The choice is based on its size, location, speed of growth, and whether urgent pressure relief is needed.

Kidney angiomyolipomas may be monitored, treated with an mTOR inhibitor, or managed with a procedure that blocks their blood supply. Kidney-preserving approaches are preferred when possible.

Developmental, educational, behavioural, and mental health support is also part of treatment. Assessments can identify difficulties with language, learning, attention, anxiety, sleep, or social communication. Support should be matched to the person's needs rather than based only on an intelligence score.

Complications

Seizures may become frequent or difficult to control. Rarely, a prolonged seizure develops into status epilepticus, which is a medical emergency.

A growing SEGA can block the normal movement of fluid through the brain. This may cause hydrocephalus, meaning a buildup of fluid and pressure. Possible signs include worsening headache, repeated vomiting, unusual sleepiness, vision changes, or new weakness.

Kidney angiomyolipomas contain abnormal blood vessels that can bleed. Kidney cysts or repeated procedures may also reduce kidney function over time. High blood pressure can occur and may not cause noticeable symptoms.

LAM can gradually damage lung tissue. It also raises the risk of a collapsed lung, which may cause sudden chest pain and shortness of breath.

Developmental, behavioural, and mental health effects are sometimes grouped under the term TSC-associated neuropsychiatric disorders , or TAND. These effects can influence education, work, relationships, and independence. They may change over time and can occur even when seizures are controlled.

Prevention

Tuberous sclerosis-1 cannot be prevented through diet, exercise, vaccination, or choices made during pregnancy. It is caused by a genetic change that is inherited or arises by chance.

Genetic counselling can explain inheritance, testing for relatives, and reproductive options. It cannot predict exactly how mildly or severely an affected child would experience the condition.

Although the condition itself cannot be prevented, regular monitoring can reduce the chance that a growing lesion or organ problem goes unnoticed. Following the recommended schedule for scans, blood pressure checks, kidney tests, seizure review, and developmental assessment can support earlier treatment.

Living with it and what to expect

Tuberous sclerosis-1 changes across life. Heart growths found in infancy often shrink, while kidney or lung changes may become more important in adulthood. Skin findings may become more visible with age. Brain lesions usually remain noncancerous, although some can grow or cause seizures.

Regular follow-up continues even when you feel well. Kidney growths, high blood pressure, or a SEGA may not cause early symptoms. Adults who had few childhood problems may still need kidney and lung surveillance.

School, work, and independence differ from person to person. Some people have typical learning and live independently. Others need educational adjustments, help with daily activities, or long-term caregiving. Anxiety, attention problems, sleep disturbance, and social difficulties deserve assessment rather than being dismissed as unavoidable parts of the condition.

Pregnancy may require planning with specialists familiar with tuberous sclerosis. Kidney, seizure, heart, and lung health may need review, and some medicines may need to be reconsidered before pregnancy.

Many people with TSC1-related tuberous sclerosis lead active adult lives. Long-term health is influenced by seizure control and the extent of kidney, brain, and lung involvement. Lifelong, coordinated follow-up helps identify changes early, but it cannot guarantee a particular outcome.

When to get medical help

Call emergency services for:

Use the person's seizure emergency plan when one is available. Do not restrain them or place anything in their mouth.

Seek prompt medical advice for a first suspected seizure, a clear change in seizure pattern, new shortness of breath, visible blood in urine, persistent headache, unexplained loss of skills, or major changes in mood or behaviour.

Routine follow-up is appropriate for stable skin changes, planned surveillance scans, medication monitoring, developmental reviews, and discussion of genetic testing or pregnancy planning. Do not wait for a routine appointment if symptoms are new, rapidly worsening, or severe.

- A seizure lasting five minutes or longer
- Repeated seizures without recovery between them
- Serious breathing difficulty or sudden chest pain with breathlessness
- Sudden severe abdominal or side pain with fainting, weakness, or blood in urine
- Severe headache with repeated vomiting, unusual drowsiness, confusion, or new weakness
- Immediate risk of self-harm or suicide

Common questions

Is tuberous sclerosis-1 cancer?

No. Most growths caused by tuberous sclerosis are noncancerous. They can still cause harm by bleeding, disrupting organ function, or pressing on nearby structures. Some kidney cancers can occur, so unusual kidney findings need specialist assessment.

What is the difference between TSC1 and TSC2?

They are different genes involved in the same cell-growth pathway. A harmful change in either

gene can cause tuberous sclerosis complex. TSC1-related cases are often milder on average, but there is wide overlap and the gene alone cannot predict an individual's needs.

Can I have tuberous sclerosis-1 without seizures?

Yes. Seizures are common, but not everyone has them. Some people are diagnosed after skin findings, kidney imaging, a heart finding, or genetic testing prompted by an affected relative.

Can tuberous sclerosis-1 skip a generation?

A person who inherits a disease-causing TSC1 variant will usually have some features, although they may be very mild. It may appear to skip a generation if signs were overlooked or if mosaicism makes them difficult to detect.

Will the growths keep getting bigger?

Not all of them do. Heart rhabdomyomas often shrink, while kidney angiomyolipomas or certain brain growths may enlarge. Regular imaging is used because growth cannot be judged from symptoms alone.

Is there a cure for tuberous sclerosis-1?

There is no cure that removes the genetic change. Treatment can control seizures, shrink some growths, protect organ function, and support development and mental health.

Sources

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