CONDITION

Phakomatosis TS

Phakomatosis TS is a genetic condition that causes noncancerous growths in several 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, kidney growths, learning differences

WHAT CAUSES IT
Inherited or new TSC1 or TSC2 gene change

HOW IT IS DIAGNOSED
Clinical criteria and genetic testing

MAIN TREATMENT
Monitoring, medicines, procedures, and surgery

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 lead active lives with lifelong monitoring

Overview

Phakomatosis TS is a term used for tuberous sclerosis complex , often shortened to TSC. It is a lifelong genetic condition that can affect several parts of the body. Its effects differ greatly from one person to another, even within the same family.

TSC causes cells to grow in an unregulated way. This can produce growths called hamartomas in the brain, skin, kidneys, heart, lungs, or eyes. Hamartomas are made from tissue normally found in that organ. Most are not cancer, but their size or location can still cause health problems.

The brain is commonly affected. Some people develop seizures, including seizures beginning in infancy. TSC can also affect learning, sleep, attention, mood, communication, and behavior. Together, these are sometimes called TSC-associated neuropsychiatric disorders, or TAND.

Regular monitoring matters because some changes cause few symptoms at first. Finding a growing kidney or brain lesion early may allow treatment before it causes lasting harm.

Symptoms

TSC may be recognized in infancy, childhood, or adulthood. Some people have many features, while others have only a few.

Skin findings are common and may include:

Pale patches may be easier to see under a special ultraviolet lamp. They can also occur in people without TSC, so they do not confirm the condition by themselves.

Brain-related features include different types of seizures, developmental delay, intellectual disability, headaches, or changes in movement and balance. Infantile spasms are brief seizures that may look like repeated head drops, body crunching, or sudden stiffening. They need prompt assessment.

Learning and behavior effects can include autism-related traits, attention difficulties, anxiety, low mood, sleep problems, aggression, or trouble with memory and planning. Intelligence can be within the usual range even when specific learning or mental health support is needed.

Kidney growths called angiomyolipomas contain blood vessels, muscle, and fat. They may not cause symptoms, but larger growths can bleed or affect kidney function. Kidney cysts can also occur.

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

Heart growths called rhabdomyomas are often found before birth or during infancy. Many shrink over time. Eye lesions usually do not affect sight, although their location and size matter.

- Pale patches called hypomelanotic macules
- Small reddish bumps on the face called angiofibromas
- Thickened, rough skin called a shagreen patch
- Growths around or beneath the nails
- A thickened patch on the forehead or scalp

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. When one is not working properly, certain cells can grow and form TSC-related lesions.

The condition 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 number and severity of features cannot be predicted from inheritance alone.

In many people, the gene change is new and was not inherited from either parent. This happens around conception or during early development. It is not caused by anything a parent did during pregnancy.

Some people have mosaic TSC , meaning the gene change is present in only some of their cells. Mosaicism may produce milder or less typical features and can make genetic testing more difficult.

Diagnosis

Diagnosis may begin after seizures, characteristic skin findings, a heart growth seen on prenatal imaging, or kidney or brain abnormalities.

A clinician will review your medical and family history and examine your skin, nails, teeth, eyes, and nervous system. Tests may include:

TSC has formal clinical diagnostic criteria based on combinations of major and minor features. A disease-causing change in TSC1 or TSC2 can also establish the diagnosis.

A negative genetic test does not always rule out TSC. Current testing may miss some changes, especially when mosaicism is present. Clinical findings remain important.

Genetic counseling can help you understand test results, inheritance, and what the findings may mean for relatives.

- MRI of the brain to look for characteristic lesions
- Kidney MRI or another appropriate imaging test
- Heart ultrasound and an electrocardiogram
- Eye examination
- Lung testing and chest imaging when relevant
- Developmental, learning, behavioral, and mental health assessments

Treatment and management

There is no treatment that removes the underlying genetic change. Management focuses on controlling symptoms, protecting organ function, and treating lesions that are growing or causing problems.

Seizures are treated with antiseizure medicines. Infantile spasms require rapid specialist assessment because early treatment is important for development. If medicines do not control seizures, options may include epilepsy surgery, dietary therapy, or devices that help regulate brain activity.

Medicines called mTOR inhibitors , including everolimus and sirolimus, act on the cell-growth pathway affected by TSC. They may be used for certain growing brain or kidney lesions, some seizures, LAM, or troublesome skin lesions. These medicines require monitoring because they can cause side effects and interact with other treatments.

A growing brain lesion called a subependymal giant cell astrocytoma, or SEGA, may be treated with an mTOR inhibitor or surgery. The choice is based on its size, growth, symptoms, and whether it is blocking the normal flow of fluid around the brain.

Kidney angiomyolipomas may be monitored, treated with an mTOR inhibitor, or managed with a procedure to block their blood supply. Surgery is sometimes needed. Care aims to control bleeding while preserving as much kidney tissue as possible.

Skin treatments can include topical medicine, laser treatment, or minor procedures. These may reduce bleeding, irritation, or visible changes, but new lesions can develop.

Developmental, educational, behavioral, sleep, and mental health needs deserve the same attention as physical findings. Support may include speech or occupational therapy, school adjustments, psychological treatment, or appropriate medication.

Lifelong monitoring usually includes brain and kidney imaging, blood pressure and kidney checks, skin and dental care, eye examinations, seizure review, and assessment of learning and mental health. Lung monitoring is particularly important for adult women and anyone with breathing symptoms.

Complications

Many TSC-related growths remain stable and cause little difficulty. Possible complications depend on which organs are involved.

Brain lesions may contribute to seizures or, less commonly, block fluid flow and raise pressure inside the skull. Warning signs can include worsening headaches, vomiting, unusual sleepiness,

vision changes, or a decline in balance.

A kidney angiomyolipoma can bleed. Sudden severe pain in the side or abdomen, weakness, fainting, or blood in the urine needs urgent assessment. Long-term kidney damage and high blood pressure can also occur.

LAM can gradually reduce lung function. Air leaking from the lung can cause a collapsed lung, with sudden chest pain and breathlessness.

Seizures, sleep disruption, learning difficulties, anxiety, depression, and behavior changes can affect school, work, relationships, and independence. These effects are not simply personal or parenting failures. They are recognized parts of TSC and may respond to targeted support.

Prevention

TSC itself cannot usually be prevented. It results from an inherited or newly occurring genetic change, not from diet, activity, infection, or choices made during pregnancy.

Regular surveillance cannot stop the condition from existing, but it can reduce preventable harm. Monitoring may find growing lesions, high blood pressure, kidney problems, or developmental needs before they become more difficult to manage.

If you have TSC and are considering pregnancy, genetic counseling can explain the chance of passing it on and discuss reproductive testing options. A medication review is also important because some TSC treatments require special planning around pregnancy.

Avoiding smoking may be especially important for people with lung involvement, although it does not prevent the genetic condition.

Living with it and what to expect

TSC changes over time. Heart growths found in babies often become smaller, while skin, kidney, or lung changes may become more noticeable later. Seizures may improve, remain stable, or require repeated treatment adjustments.

Development is also individual. Some children need extensive lifelong support. Others attend mainstream school, work, form relationships, and live independently. A person can have average intelligence while still needing help with attention, anxiety, social communication, or planning.

Routine appointments may continue even when you feel well. Brain and kidney lesions can grow without causing early symptoms, so imaging and other checks remain important. The

type and timing of follow-up are based on age, previous findings, symptoms, and treatment.

Keep an up-to-date list of medicines, previous scans, procedures, allergies, and specialist contacts. If possible, care through a TSC clinic can help coordinate neurology, kidney, lung, skin, heart, eye, genetic, and mental health services.

New symptoms should not automatically be assumed to come from TSC. Common health problems still occur and may need their own assessment.

When to get medical help

Arrange routine or prompt medical follow-up if you notice new seizures, increasing seizure frequency, changes in learning or behavior, persistent headaches, blood in the urine, worsening skin lesions, or gradually increasing breathlessness.

Seek urgent medical advice for:

Call emergency services for a seizure lasting five minutes or longer, repeated seizures without recovery, serious injury during a seizure, or trouble breathing afterward. Follow an existing emergency seizure plan if one has been provided.

Sudden chest pain with shortness of breath is also an emergency because it may indicate a collapsed lung. Severe abdominal or side pain with faintness may signal internal bleeding from a kidney lesion.

- A first suspected seizure
- Repeated infant head drops, body crunches, or sudden stiffening
- Sudden severe pain in the side or abdomen
- New weakness, fainting, or visible blood in the urine
- Persistent vomiting with headache or unusual sleepiness
- A clear decline in balance, vision, movement, or awareness

Common questions

Is phakomatosis TS the same as tuberous sclerosis?

The term is used for tuberous sclerosis complex, or TSC. "Phakomatosis" is an older broad term for genetic conditions affecting the skin and nervous system. TSC can also affect the kidneys, lungs, heart, and eyes.

Are the growths cancer?

Most TSC growths are benign, meaning they are not cancer. They can still cause problems if they grow, bleed, damage an organ, or block the flow of fluid in the brain. Cancerous kidney tumors can occur but are not the typical kidney lesion in TSC.

Can someone have mild TSC?

Yes. Some people have few symptoms and are diagnosed only after a relative is tested or an unrelated scan finds a characteristic lesion. Mild current symptoms do not remove the need for monitoring because some features appear later.

Will my child inherit it?

If you have a disease-causing TSC1 or TSC2 change, each pregnancy has a 1 in 2 chance of inheriting it. The severity cannot be predicted from the parent's symptoms. A genetics service can explain family-specific results.

Can TSC symptoms get worse with age?

Some features may change with age. Heart rhabdomyomas often shrink, while kidney, skin, and lung findings may develop or progress later. Seizure and developmental outcomes follow different courses for different people.

Can people with TSC have children?

Many can. Pregnancy planning should include genetic counseling and a review of medicines, kidney lesions, seizures, and lung health. Pregnancy may require coordinated care, particularly when LAM or significant kidney angiomyolipomas are present.

Sources

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