
Tuberous sclerosis

Summary

- Tuberous sclerosis is a genetic disorder that can target various parts of the body to varying degrees of severity.
 - Its common characteristic is the formation of tuber-like growths in the brain and perhaps other organs, including the kidneys, heart, liver and lungs.
 - These growths can cause seizures, delayed development, intellectual impairment and behaviours that seem autistic or hyperactive.
 - There is no cure for tuberous sclerosis.
-

Tuberous sclerosis is a genetic disorder that affects various parts of the body to varying degrees of severity. Its common characteristic is the formation of tuber-like growths in the brain and, perhaps, other organs, including the kidneys, heart, liver and lungs.

These growths begin to form in the brain prior to birth and interfere with brain functioning. They can cause seizures, delayed development, mental retardation, and behaviours that seem autistic or hyperactive.

With age, these growths become hard and calcified, hence the term 'sclerosis'. However, life expectancy remains normal. Around one out of two people born with tuberous sclerosis will lead normal lives with no apparent intellectual dysfunction. Estimates vary, but it is thought that approximately one in 6,000 to 10,000 people have tuberous sclerosis. There is no cure.

Causes of tuberous sclerosis

Tuberous sclerosis is caused by one of two faulty genes. In around two thirds of cases, the gene spontaneously mutates within the developing baby, for reasons unknown. This gene may also be inherited, and a person with the condition has a 50 per cent chance of passing tuberous sclerosis onto each of their children. The genes involved are on chromosome 9 (TSC1) and chromosome 16 (TSC2). An error in either of these two genes causes the proteins that regulate cell growth to malfunction.

A molecular diagnostic test for TSC is available and can be used as confirmation of a clinical diagnosis and for reproductive decision making, including pre-natal diagnosis and pre-implantation genetic diagnosis.

Symptoms of tuberous sclerosis

The symptoms of tuberous sclerosis vary from one individual to the next, depending on the severity of the condition and which areas of the body are affected. Symptoms may include:

- white patches of skin on the body
- skin rash of red pin-points across the nose and cheeks that progress to small lumps
- delayed development
- learning difficulties
- epilepsy
- autistic tendencies
- hyperactive tendencies
- sleeping difficulties.

Skin rashes symptoms

The skin rashes of tuberous sclerosis can take a range of forms, including:

- Ash leaf patch – patches of skin are white because they lack pigment. These patches tend to take the shape of a leaf and are sometimes present at birth, but usually fade over the years.
- Shagreen patch – the skin patch has a textured, raised appearance, similar to orange peel. These patches are present at birth and usually form on the lower back. They can also develop later in life.
- Facial angiofibroma – the affected person typically develops a rash across their nose and cheeks. At first, the rash appears like collections of red pin-points, then each mark develops into a small lump. When the child reaches their teenage years, little papules may also form around their finger and toenails.

Brain functioning is affected

The tuber-like growths in the brain tend to affect functioning in a variety of ways, which may include:

- Epilepsy – around 80 per cent of children with tuberous sclerosis have seizures or fits. In some cases, the epilepsy starts within a few months of birth with infantile spasms. These seizures may lessen with time and cease altogether, but medication is needed to control them as soon as possible.
- Developmental delay – depending on the severity of the condition, the child may fail to reach expected milestones within the normal age range. Whether or not a child will have developmental problems is usually apparent by their second birthday.
- Mental retardation – depending on the severity, some children may have normal intellectual function, or mild to severe retardation.
- Autistic tendencies – typical behaviours associated with autism include the tendency to avoid eye contact, and problems with language and social development.
- Hyperactive tendencies – typical behaviours associated with hyperactivity include impulsivity and overactivity.

Other organs affected

A range of other organs may be affected by the tuber-like growths, including the lungs, kidneys, heart, bones and liver. Eighty per cent of people with tuberous sclerosis have affected kidneys. Rarely, children with affected eyes may experience visual problems.

Diagnosis of tuberous sclerosis

If a child is diagnosed with tuberous sclerosis, other family members should be tested. This is to find out whether the child inherited the condition or if a spontaneous gene mutation was responsible. Tuberous sclerosis is diagnosed using a number of clinical tests, including:

- physical examination
- eye examinations, which may reveal retinal abnormalities
- tests to check for heart abnormalities
- a computed tomography (CT) scan, since growths will show as white patches in the brain
- other possibly affected organs, such as lungs and kidneys, may be checked for the presence of tuber-like growths to confirm diagnosis.

If the TSC gene mutation can be found in the person diagnosed with TSC, genetic testing may be used to test for TSC in family members. The gene mutation can be found in 70 to 90 per cent of cases. This gene test can also be used for pre-natal testing. If the gene mutation cannot be found in the person with TSC, echocardiography of the baby's heart may show the characteristic lesions.

Treatment for tuberous sclerosis

There is no cure for tuberous sclerosis. Treatment aims to ease some of the associated symptoms. Depending on the severity of the condition, treatment options may include:

- Anti-epileptic drugs – to treat the associated seizures. Medication needs to be carefully monitored to make sure the child isn't over-sedated.
- Brain surgery – if seizures cannot be controlled, it may be possible to remove lesions in the brain to reduce seizures.
- New drug treatments – some new medication is in clinical trials to treat tuberous sclerosis tumours in the brain, lungs, kidneys and skin.
- Skin treatments – the advice of a dermatologist should be sought to treat the skin rashes. Laser therapy can

remove the papules.

- Occupational therapy – can help the child acquire coping skills and strategies.
- Speech therapy – can assist communication skills.
- Stimulation – can encourage the child’s social and intellectual development. Options could include playgroups and home teaching.

Where to get help

- Your doctor
- Neurologist
- Paediatrician
- Dermatologist
- Occupational therapist
- Australasian Tuberous Sclerosis Society Tel. 1300 733 435

Things to remember

- Tuberous sclerosis is a genetic disorder that can target various parts of the body to varying degrees of severity.
- Its common characteristic is the formation of tuber-like growths in the brain and perhaps other organs, including the kidneys, heart, liver and lungs.
- These growths can cause seizures, delayed development, intellectual impairment and behaviours that seem autistic or hyperactive.
- There is no cure for tuberous sclerosis.

This page has been produced in consultation with and approved by:

Australasian Tuberous Sclerosis Society

Content on this website is provided for information purposes only. Information about a therapy, service, product or treatment does not in any way endorse or support such therapy, service, product or treatment and is not intended to replace advice from your doctor or other registered health professional. The information and materials contained on this website are not intended to constitute a comprehensive guide concerning all aspects of the therapy, product or treatment described on the website. All users are urged to always seek advice from a registered health care professional for diagnosis and answers to their medical questions and to ascertain whether the particular therapy, service, product or treatment described on the website is suitable in their circumstances. The State of Victoria and the Department of Health & Human Services shall not bear any liability for reliance by any user on the materials contained on this website.

For the latest updates and more information, visit www.betterhealth.vic.gov.au

Copyright © 1999/2019 State of Victoria. Reproduced from the Better Health Channel (www.betterhealth.vic.gov.au) at no cost with permission of the Victorian Minister for Health. Unauthorised reproduction and other uses comprised in the copyright are prohibited without permission.