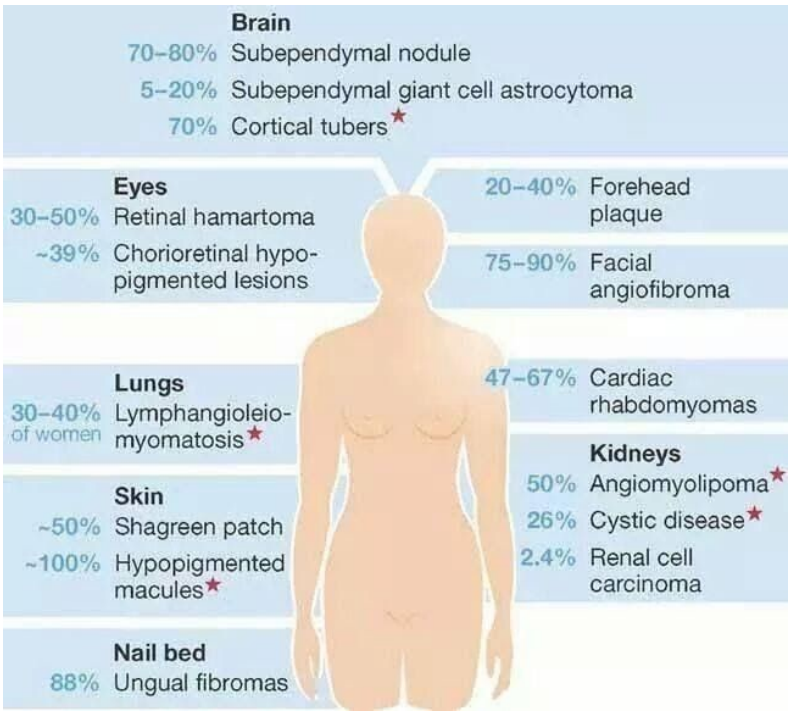
Tuberous Sclerosis

Overview

Tuberous sclerosis complex (TSC) affects different parts of the body to varying degrees of severity. Its common characteristic is the formation of non-cancerous tumours in the brain and other organs, including the kidneys, heart, liver and lungs.

TSC growths begin to form in the brain prior to birth and others can begin to grow later in life. While TSC can cause seizures, delayed development, intellectual disability and challenging behaviours, each person with TSC can show different symptoms.



Many people born with TSC will lead normal lives. It is thought that about one in 6,000 to 10,000 people have TSC. While there is no cure for TSC, there are new treatment options and lifelong, regular surveillance can help to protect the health of people with TSC.

Tuberous sclerosis is a genetic condition

Tuberous sclerosis is caused by changes in one of two genes called TSC1 and TSC2. A change in either of these genes can cause uncontrolled cell growth.

In two thirds of cases, there is no family history of the condition and the genetic change arises randomly in an individual. The genetic change can also be inherited, and a person with TSC has a 50 per cent chance of passing it on to each of their children.

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
- autism
- hyperactive tendencies
- sleeping difficulties.

Skin symptoms of tuberous sclerosis

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
- shagreen patch – the skin patch has a textured, raised appearance, similar to orange peel. These patches can be present at birth and usually form on the lower back. They can 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, papules may also form around their finger and toenails.

Brain functioning is affected by tuberous sclerosis

TSC can affect the brain in a variety of ways, including:

- **epilepsy** – around 80 per cent of people 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 not reach expected milestones within the normal age range. Whether or not a child will have developmental problems with TSC is usually apparent by their second birthday
- intellectual disability – depending on the severity, some children may have normal intellectual function, or mild to severe intellectual disability
- **autism spectrum disorder (ASD)** – typical behaviours associated with autism include the tendency to avoid eye contact, and problems with communication and social development

- mental health issues – many people with TSC experience anxiety disorders and depression.

Other organs affected by tuberous sclerosis

Any organ of the body can be affected by TSC tumours, including the lungs, kidneys, heart, bones and liver. Eighty per cent of people with TSC have affected kidneys. Rarely, children with affected eyes may experience visual problems.

Diagnosis of tuberous sclerosis

Tuberous sclerosis is diagnosed using a number of clinical tests, including:

- physical examination
- imaging of various parts of the body through MRI, CT or ultrasound
- eye examinations, which may reveal retinal abnormalities
- tests to check for heart abnormalities.

A genetic test for TSC is available and can be used to confirm a clinical diagnosis. The genetic variant can be found in 70 to 90 per cent of cases. If the genetic variant is found in an individual with TSC, genetic testing can be offered to family members. If the genetic variant is not found in the person with TSC, this does not mean that they do not have TSC. Instead, clinical tests can be used.

For women or couples with a known TSC genetic variant who wish to test their baby for TSC before birth, there are two main testing options:

- prenatal diagnostic testing – can check for the genetic change and will show if the baby has TSC. This can be done though **chorionic villus sampling** or **amniocentesis**
- IVF with pre-implantation genetic diagnosis (PGD) – for women undergoing IVF, using IVF technology to test embryos for the genetic change before embryo implantation and pregnancy.

Treatment for tuberous sclerosis

There is no cure for tuberous sclerosis but there are a variety of treatments that can be used for the various signs and symptoms of TSC. Treatment options may include:

- mTOR inhibitor medicines – can be used to treat brain tumours, kidney tumours and epilepsy caused by TSC
- anti-epileptic medicines – 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
- skin treatments – dermatologists can provide advice on both surgical and medical treatment for the skin signs of TSC, including mTOR inhibitor creams.
- occupational therapy – can help children acquire skills and strategies
- speech therapy – can assist communication skills.