What causes TS?

TS is caused by an alteration in a gene. Genes are the unique instructions which make each of us an individual. There are many thousands of different genes; each gene is the coded instruction for the creation of a specific enzyme or other protein.

We know of two genes that are associated with TS, called TSC1 and TSC2. Affected people have an alteration (‘mutation’) in either TSC1 or TSC2. This stops the gene from working properly.

About 70 per cent of people with TS are the first in their family to be affected. This is because the alteration in the gene has arisen in them for the first time.

The remaining 30 per cent of people with TS will have inherited it from a parent. Sometimes the parent may be very mildly affected, to the extent that they are not aware that they have TS. Parents of affected children should be carefully examined for signs of TS, and if they are planning further children, gene testing may be carried out.

If a person affected with TS has children each child has a one in two chance of inheriting the altered gene.

Is there a blood test for TS?

It is now possible to analyse TSC1 and TSC2 to find the gene alteration. The test detects an alteration in most, but not all, affected people.

Gene testing is not necessary in everyone but can be offered to people where their diagnosis is uncertain, to people who are at risk in a family or to those who might wish to have prenatal diagnosis (testing a pregnancy). Prenatal diagnosis and family testing can only be offered if the gene alteration has been identified in an affected family member.

More information

Tuberous Sclerosis Association
CAN Mezzanine, 32-36 Loman Street,
London SE1 0EH
Tel: 020 7922 7731
E-mail: admin@tuberous-sclerosis.org
www.tuberous-sclerosis.org

Feedback

We appreciate and encourage feedback. If you need advice or are concerned about any aspect of care or treatment please speak to a member of staff or contact the Patient Advice and Liaison Service (PALS):

Freephone: 0800 183 0204
From a mobile or abroad: 0115 924 9924 ext 65412 or 62301
E-mail: pals@nuh.nhs.uk
Letter: NUH NHS Trust, c/o PALS,
Freepost NEA 14614,
Nottingham NG7 1BR

www.nuh.nhs.uk

The Trust endeavours to ensure that the information given here is accurate and impartial.

Adapted from leaflet by Clinical Genetics at Guy’s and St Thomas’ NHS Foundation Trust.

Dr Jacqueline Eason, Clinical Genetics © October 2018 All rights reserved. Nottingham University Hospitals NHS Trust. Review October 2020. Ref: 1035/v3/1018/AH.
What is tuberous sclerosis (TS)?
Tuberous sclerosis (TS), also known as tuberous sclerosis complex (TSC), is a genetic condition that affects many different systems in the body. Its effects vary greatly between individuals, even within the same family, with some people being so mildly affected they don’t know they have it, while others have major difficulties from early life.

TS derives its name from the ‘tuber’ like growths that are seen in the brain. The growths are benign (not cancerous) and they become hard with age (sclerotic). They can be seen as small white patches on a brain scan. Other benign growths can also affect many other organs, particularly the skin, eyes, heart, kidneys and lungs. Often these do not cause any problems.

TS affects about one in 7,000 people. This means that there are approximately 8,000 affected people in the UK.

How is TS diagnosed?
TS may be diagnosed at any time throughout life depending on the severity of the symptoms. There are a number of different signs of TS, and a combination of these is necessary to make the diagnosis. They include:

**Skin** - white skin patches may be present from birth; there may be a characteristic facial rash (facial angiofibroma) across the nose and cheeks, often appearing during childhood; small lumps of skin (fibromas) around the finger or toe nails may appear later in childhood or adolescence. A fleshy lump is often found on the lower back (shagreen patch).

**Epilepsy** - seizures occur in about 70 per cent of people with TS. They may start at any time, but typically in childhood and often during the first year. Babies may have infantile spasms (also known as salaam attacks) that need to be treated promptly. People with TS may have different types of seizures at different times of their lives and seizures sometimes cease altogether.

**Kidneys** - about 70 to 80 per cent of people with TS will have kidney involvement. Occasionally multiple cysts in the kidneys are detected in a baby during a routine antenatal ultrasound scan or soon after birth. However, in most people, benign tumours (angiomyolipoma) are detectable later in childhood or adulthood. These often do not cause problems, but can sometimes bleed and should be monitored. Very occasionally, other tumours of the kidney may develop.

**Heart** - benign heart tumours (cardiac rhabdomyomas) are an early sign of TS which are seen in about 60 per cent of children. They may be detected before birth on a routine ultrasound scan. They rarely cause any problems, and usually disappear.

**Development delay** - about 40 to 50 per cent of people with TS have normal intelligence, but the remainder have learning difficulties which vary from mild to severe.

**Lungs** - Lymphangioleiomyomatosis (LAM) occurs in approximately 30% of ladies with TS but only causes breathing problems in a small number. Assessment for LAM should be undertaken once ladies with TS reach adulthood.

**Behaviour** - Behavioural problems are common. About 25 per cent of people with TS have autism, and another 25 per cent show some autistic features. Attention deficit disorder and hyperactivity are common in childhood, and anxiety, paranoia and depression are more common in adults. Sleep disturbance is also seen more commonly in people with TS.

What treatment is available?
Unfortunately there is no cure for TS. However, many of the different aspects of the disorder can be treated. Newly developed treatments mean that routine screening particularly for kidney problems is recommended.

Treatment will involve a number of different professionals, depending on the symptoms. For example, epilepsy should be managed by a neurologist or paediatrician experienced in the management of epilepsy.

Skin problems can often be treated and would need to be referred to a dermatologist (skin specialist). Kidney problems may require the input of a kidney specialist.

What is the outlook?
TS is a variable condition, so that the long term outlook depends on the symptoms and severity of the disorder in each individual.

About 50 per cent of people will be intellectually normal and lead normal lives. Others will have a degree of intellectual impairment, but many of these people will have a normal lifespan.