

What causes TS?

TS is caused by an alteration in a gene. A gene is a segment of DNA that has a particular purpose. A gene codes for (contains the chemical information necessary for the creation of) a specific enzyme or other protein. Genes determine our personal characteristics such as eye colour and hair colour. We know of two genes that can result in TS, named TSC1 and TSC2. Affected people are found to have an alteration in *either* TSC1 or TSC2. About 70% 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. However, when they have children each child has a 50:50 chance of inheriting the altered gene. The remaining 30% 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 are affected. Parents of affected children should be carefully examined for signs of TS, and if they are planning further children, gene testing may be performed (see below).

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 all people but can be offered to those in whom the diagnosis is uncertain, or to people who are at risk in a family or to those who might wish to have prenatal diagnosis (a test on a pregnancy). Prenatal diagnosis and family testing can only be offered if the gene alteration has been identified in an affected family member.

For more information about Tuberous Sclerosis:

Clinical Genetics Departments

Northern Scotland (main base Aberdeen)
Tel: 01224 552120 Fax: 01224 559390
(Aberdeenshire, Moray, Highland, Western & Northern Isles)

Tayside (main base Dundee)
Tel: 01382 632035 Fax: 01382 645731
(Perth & Kinross, Angus, North East Fife)

South East Scotland (main base Edinburgh)
Tel: 0131 651 1012 Fax: 0131 651 1013
(Borders, Lothian, South West Fife)

West of Scotland (main base Glasgow)
Tel: 0141 201 0808 Fax: 0141 201 0361
(Glasgow, Argyll & Bute, Ayrshire, Dumfries & Galloway, Stirling, Lanarkshire, Falkirk)

The Tuberous Sclerosis Association (TSA) provides information and support for individuals with TS and their families, you can contact them at the following address:

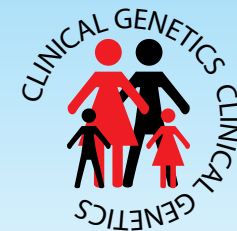
The Tuberous Sclerosis Association (TSA)

Mrs Janet Medcalf
Head of Support Services
PO Box 9644
Bromsgrove
B61 0FP
Email: support@tuberous-sclerosis.org

Seen in clinic by.....

This leaflet is based on information provided by the TSA and was written by Guys & St Thomas Clinical Genetics & updated by Genetic Interest Group Scotland. Last updated Feb 2002.

Tuberous Sclerosis (TS)



A patient
information
leaflet

Introduction

Tuberous sclerosis (TS) is also known as tuberous sclerosis complex (TSC) because the condition affects many different systems of the body. It is highly variable 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.

How common is TS?

TS is thought to affect about 1 in 7000 people. This means that there are approximately 8000 affected people in the UK alone.

What is TS?

TS derives its name from the "tuber"-like growths that are seen in the brain. The growths are benign (not cancerous). These become hard with age (sclerotic) and can be seen as small white patches on a brain scan.

Other benign growths can affect many other organs of the body, particularly the skin, eyes, heart, kidneys and lungs. Often these do not cause any problems.

How is it 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 some of these signs is necessary to make the diagnosis. These include:

Skin

White skin patches that may be present from birth. Characteristic facial rash (facial angiofibroma) across the nose and cheeks. This often appears during childhood.

Small lumps of skin (fibromas) around the finger or toe nails. These may appear later in childhood/adolescence.

A fleshy lump often found on the lower back (shagreen patch).

Epilepsy

Seizures appear in about 70% of people with TS. They may start at any time, but typically start in childhood, 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.

Developmental delay

About 40-50% of people with TS have normal intelligence, but the remainder have learning difficulties that vary from mild to severe.

Kidneys

About 70-80% of people with TS will have kidney involvement. Occasionally multiple cysts in the kidneys are detected in a baby on routine antenatal ultrasound scan, or soon after birth. However, in most people, kidney tumours (angiomyolipoma) are detectable later in childhood or adulthood. These frequently do not cause problems, but can sometimes bleed and therefore need to be monitored. Very occasionally, malignant tumours of the kidney may develop.

Heart

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

Behaviour

Behavioural problems are common. About 25% of people with TS are autistic, and another 25% 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. Treatment will involve a number of different professionals, depending on symptoms. For example, epilepsy should be managed by a neurologist or paediatrician experienced in the management of epilepsy. The skin problems often can be treated and would need referral 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 symptoms and severity of the disorder in any individual. About 50% 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.