

# Tuberous Sclerosis

## Information Guide

### 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 difficulty from early life.

### How common is TS?

TS is thought to affect about 1 in 7,000 people. This means that there are approximately 8,000 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 7 to 10 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 seizure 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 a quarter of people with TS are autistic and another quarter 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 half the people with TS will be intellectually normal and will lead normal lives. Others will have a degree of intellectual impairment, but many of these people will have a normal lifespan.

### **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 7 out of 10 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 3 out of 10 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 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, 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 additional information or support

If you have any further questions about tuberous sclerosis, please contact:

### Genetic Medicine

6<sup>th</sup> Floor, St Mary's Hospital, Oxford Road, Manchester M13 9WL

Telephone 0161 276 6506

Fax 0161 276 6145

Department staffed Monday–Friday, 9.00am to 5.00pm.

The Tuberous Sclerosis Association (TSA) provides information and support for individuals with TS and their families:

### The Tuberous Sclerosis Association (TSA)

Mrs Janet Medcalf, Head of Support Services,

PO Box 9644, Bromsgrove B61 0FP

Email [support@tuberous-sclerosis.org](mailto:support@tuberous-sclerosis.org)

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