

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 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 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.

Translation and interpretation service

Do you have difficulty speaking or understanding English ?

আপনি কি ইংরেজীতে বুঝতে কিংবা বুঝাতে পেরেছেন ? (BENGALI)

क्या आपको अंग्रेजी बोलने या समझने में कठिनाई है ? (HINDI)

તમે ભાષા કારણે વાતચીત કરવામાં મુશ્કેલી આવે છે ? (GUJARATI)

ਕਿ ਤੁਹਾਨੂੰ ਅੰਗ੍ਰੇਜ਼ੀ ਬੋਲਣ ਜਾਂ ਸਮਝਣ ਵਿਚ ਦਿੱਕਤ ਹੈ ? (PUNJABI)

Miyey ku adagtahay inaad ku hadasho Ingriisida aad sahamto (SOMALI)

هل لديك مشاكل في فهم اوالتكلم باللغة الانجليزية ؟ (ARABIC)

你有困難講英語或明白英語嗎？ (CANTONESE)

کیا آپ کو انگریزی سمجھنے اور سمجھانے میں دقت پیش آتی ہے؟ (URDU)

☎ Telephone Number 0161 276 6202/6342

This leaflet is based on information provided by the TSA.

We would like to acknowledge our Clinical Genetics colleagues at Guy's and St Thomas' Hospital NHS Trust who wrote the original version of this leaflet.

Please let us know if you would like this leaflet in another format (e.g. large print, Braille, audio).

Seen in clinic by(doctor)

and(genetic counsellor)

Telephone number

Family reference number

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Tuberous Sclerosis (TS)

Information for patients



INVESTOR IN PEOPLE



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 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:

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

Smoking

Smoking is only permitted within the external designated areas. You may be putting others at risk if you smoke in non-designated areas. The Manchester Stop Smoking Service can be contacted on Tel: (0161) 205 5998 (www.stopsmokingmanchester.co.uk).

Suggestions, concerns and complaints

If you wish to make a comment, have a concern or want to complain, it is best in the first instance to speak to the manager of the ward or department involved.

The Trust has a Patient Advice and Liaison Service (PALS) who can be contacted on (0161) 276 8686 and via e-mail: pals@cmmc.nhs.uk. They will help you if you have a concern, want advice or wish to make a comment or complaint.

Information leaflets about the service are readily available throughout the Trust. Please ask any member of staff for a copy.

For more information

If you need more advice about any aspect of Tuberous Sclerosis, you are welcome to contact:

Genetic Medicine
Sixth Floor
Saint Mary's Hospital
Oxford Road
Manchester, M13 9WL
Telephone: 0161 276 6506 Reception
Facsimile: 0161 276 6145

Department staffed Monday - Friday 9.00 am to 5.00 pm

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 out of 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 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.