Saint Mary’s Hospital
Manchester Centre for Genomic Medicine

Tuberous Sclerosis Complex (TSC)
Information For Patients
Introduction

Tuberous sclerosis complex (TSC) is also known as tuberous sclerosis (TS). TSC affects many different systems of the body such as the brain, skin and kidneys. Individuals with TSC, even within the same family, can experience different problems. Some people have such minor problems that they don’t know they have TSC, while others have major difficulties from early life.

Why is it called TSC?

TSC derives its name from the ‘tuber’-like growths that may be 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 common is TSC?

TSC is thought to affect about 1 in 7,000 people. This means that there are approximately 8,000 affected people in the UK alone.

How is it diagnosed?

TSC may be diagnosed at any time throughout life depending on the severity of the symptoms. There are a number of different signs of TSC and a combination of some of these signs is necessary to make the diagnosis. A diagnosis can usually be made after an examination by a doctor and scans such as an ultrasound of the kidneys. Sometimes a genetic blood test is possible.
Signs and symptoms of TSC include:

**Skin**

Virtually all people with TSC will have some of the following skin features. They do not cause any serious medical problems.

- White skin patches (hypomelanotic macules). These may be present from birth.
- Characteristic facial rash (facial angiofibroma). This may appear across the face and cheeks, often during childhood.
- Small lumps of skin (fibromas) around the finger or toe nails. These may appear later in childhood/adolescence.
- A fleshy lump may be found on the lower back (shagreen patch).

**Epilepsy**

About 7 out of 10 (70%) people with TSC will have fits (epilepsy). 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 TSC may have different types of seizures at different times of their lives with seizures sometimes stopping altogether.

**Developmental delay**

About 4-5 out of 10 (40-50%) of people with TSC have normal intelligence, but the remainder have learning difficulties that vary from mild to severe.

**Behaviour**

Behaviour problems are common. About 1 in 4 (25%) people with TSC are autistic and another 1 in 4 (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 TSC.
**Kidneys**

About 7-8 out of 10 (70-80%) people with TSC have kidney involvement. Occasionally multiple cysts in the kidneys are detected in a baby on routine antenatal ultrasound or soon after birth. However, in most people, kidney growths (angiomyolipoma) are detected in childhood or adulthood. These can increase in size or number over time. They usually do not cause problems, when small but if they become large they 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 TSC and are seen in about 6 out of 10 (60%) children. They may be detected before birth on a routine ultrasound scan. They rarely cause any problems, and usually disappear without any treatment. Abnormalities of heart rhythm can rarely occur.

**Eyes**

Changes in the retina at the back of the eye such as lighter patches (achromic patches) or small growths (hamartomas) can be seen on examination by an eye specialist. They occur in 3 out of 4 (75%) of people with TSC. They do not usually cause any difficulties with vision or need any treatment.

**Lungs**

The lungs can be affected by a condition called lymphangioleiomyomatosis (LAM). This can cause shortness of breath or a collapsed lung. It is rare for men with TSC to experience these problems.

**Subependymal Nodules**

These are benign brain growths that can be seen on a brain scan in 8 out of 10 (80%) people with TSC. In up to 1 in 10 people with TSC these grow to a size that may require treatment when they are called subependymal giant cell astrocytomas (SEGA).
What treatment is available?

Unfortunately there is no cure for TSC. 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. Laser therapy can be used to treat some of the skin problems. New drugs such as mTOR inhibitors are being developed. Kidney problems will require the monitoring and sometimes treatment by a nephrologist (kidney specialist). All children with TSC should have a developmental and behavioural assessment by a paediatrician.

What is the outlook?

TSC 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 TSC are able to lead normal lives. Others will have a degree of intellectual impairment, problematic epilepsy and/or insufficient kidney function.

What causes TSC?

TSC is caused by an alteration in a gene. DNA is our genetic information and consists of several thousand genes. Each gene is an instruction for a particular task. Genes determine our personal characteristics such as eye colour and hair colour. We know of two genes that can result in TSC when changed, and these are called TSC1 and TSC2.

About 2 out of 3 people with TSC are the first in their family to have TSC. This is because the alteration in the gene happened in them for the first time. However, when they themselves have children each child has a 1 in 2 (50%) chance of inheriting the altered gene. The remaining 1 out of 3 people with TSC will have inherited TSC from a parent. Sometimes the parent has such minor problems that they are not aware that they have TSC. Parents of children should be carefully examined for signs of TSC as they may need medical checks themselves if they also have TSC.
Is there a blood test for TSC?

It is now possible to analyse TSC1 and TSC2 to try to find the gene alteration. The test detects an alteration in about 8 out of 10 (80%) people with TSC. 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).

Pregnancy

Additional ultrasounds in pregnancy are available to monitor the baby’s heart, brain and kidneys. Prenatal diagnosis and family testing can only be offered if the gene alteration has been identified in a family member who has TSC.

For more information

If you need more advice about any aspect of tuberous sclerosis complex, you are welcome to contact:

**Manchester Centre for Genomic 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

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

The Tuberous Sclerosis Association (TSA)  
CAN Mezzanine  
32-36 Loman Street  
LONDON, SE1 0EH

Christine Beal - TSC Adviser (North East and North West England and Wales) 01924 849937  
christine.beal@tuberous-sclerosis.org
Suggestions, concerns and complaints

If you would like to provide feedback you can:

• Ask to speak to the ward or department manager.

• Write to us: Patient Advice and Liaison Services, 1st Floor, Cobbett House, Manchester Royal Infirmary, Oxford Road, Manchester  M13 9WL

• Log onto the NHS Choices website www.nhs.uk - click on ‘Comments’.

If you would like to discuss a concern or make a complaint:

• Ask to speak to the ward or department manager – they may be able to help straight away.

• Contact our Patient Advice and Liaison Service (PALS) – Tel: 0161 276 8686 e-mail: pals@cmft.nhs.uk. Ask for our information leaflet.

We welcome your feedback so we can continue to improve our services.
No Smoking Policy

The NHS has a responsibility for the nation’s health.

Protect yourself, patients, visitors and staff by adhering to our no smoking policy. Smoking is not permitted within any of our hospital buildings or grounds.

The Manchester Stop Smoking Service can be contacted on Tel: (0161) 205 5998 (www.stopsmokingmanchester.co.uk).

Translation and Interpretation Service

It is our policy that family, relatives or friends cannot interpret for patients. Should you require an interpreter ask a member of staff to arrange it for you.

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