Saint Mary's Hospital
Manchester Centre for Genomic Medicine

Information for Patients

Tuberous Sclerosis Complex

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.

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 a brain scan and a kidney scan. 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 and do not tan.
- Characteristic facial rash (facial angiofibromas). 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

Approximately 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

Approximately 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

Approximately 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. Most commonly, kidney growths (angiomyolipoma) are detected in childhood or adulthood. These can increase in size or number over time. When they are small they usually do not cause problems, however if they become large they can sometimes bleed and therefore may need treatment. 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. It is important to refrain from smoking and to avoid medicines that contain oestrogen.

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. For this reason, we advise people with TSC to have regular medical check-ups so treatment can be started promptly if needed. 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 or topical medicines can be used to treat some of the skin problems. Kidney problems will require monitoring and sometimes treatment will be arranged by a nephrologist (kidney specialist). New drugs such as Everolimus are now being used to treat some of the problems experienced by people with TSC.

All children with TSC should have a developmental and behavioural assessment by a paediatrician.

What is the outlook?

TSC is a variable condition, so the long term outlook depends on the 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.

Approximately 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 with TSC 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 approximately 8 out of 10 (80%) people with TSC. Gene testing is not necessary in all people.

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 TSC, you are welcome to contact:

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Saint Mary's Hospital
Oxford Road
Manchester
M13 9WL

Telephone: (0161) 276 6506 (Reception)
Facsimile: (0161) 276 6145

The department is staffed Monday–Friday, 9.00 am to 5.00 pm.

Website: www.mangen.co.uk

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)
c/o Nightingale House,
46-48 East Street,
Epsom,
Surrey
KT17 1HQ

Their website address is: www.tuberous-sclerosis.org

Telephone: 0300 222 5737 (office) 0808 801 0700 (support)

E-mail: support@tuberous-sclerosis.org