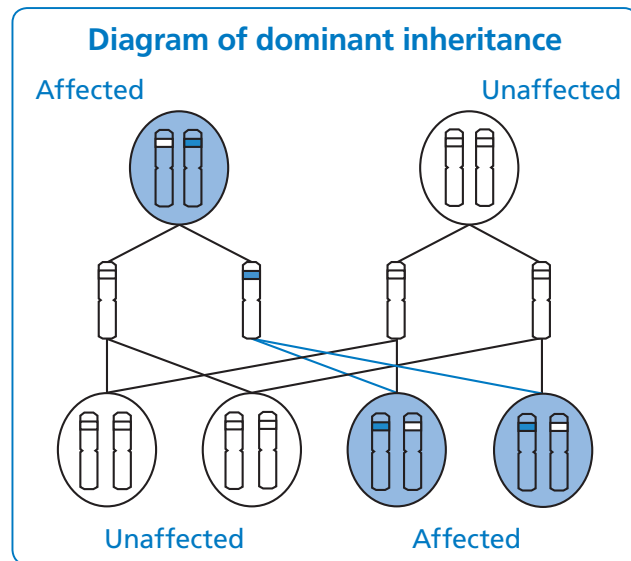


How is TSC inherited?

We all have two copies of the gene because we inherit one copy from each of our parents. The TSC gene is a dominant gene. This means that if someone inherits one faulty copy of the gene, it is likely that they will develop some symptoms of TSC, though in some cases people are not adversely affected by the condition. If someone has a faulty copy of the gene, each of their children will have a 1 in 2 (50%) chance of inheriting the faulty copy of the gene.



Planning a family

With each child a person with TSC has, there is a 1 in 2 chance that the gene alteration will be passed on. However, there is no way to determine how severely a person with the alteration will be affected by the condition. A parent who is mildly affected may have a child who is

more severely affected, or vice versa. If you are affected by TSC and are planning to start a family, you can contact your local genetics unit to discuss this further.

Further Information

For further information and support about TSC you could contact the Tuberous Sclerosis Association (TSA):

Tuberous Sclerosis Association
PO Box 12979
Barnt Green
Birmingham
B45 5AN

Email: diane.sanson@tuberous-sclerosis.org
Web: www.tuberous-sclerosis.org
Tel: 0121 445 6970

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Birmingham Women's **NHS**
NHS Foundation Trust

Tuberous Sclerosis

An information leaflet for patients and families

If you need more advice about any aspect of Tuberous Sclerosis please contact:

Clinical Genetics Unit
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NHS Foundation Trust
Mindelsohn Way
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Introduction

Tuberous sclerosis (TSC) is a condition that can affect a variety of systems within the body. It is caused by an alteration in a gene which regulates the growth of cells in the body. It results in the growth of extra, otherwise normal tissue within a number of organs such as the brain, heart, eyes, skin, lungs or kidneys. It is sometimes called Tuberous Sclerosis Complex, because of the wide variety of effects it can have.

Possible features of TSC

TSC growths can occur in various organs throughout the body, and a single affected person may only show some of these symptoms which can include:

- **Skin:** Patches of white skin (hypomelanotic patches) may be seen at birth. Rashes (angiofibromas) can develop on the face during childhood and adolescence. Other signs include patches of raised or leathery skin on the forehead or lower back, and small growths around the nails. The symptoms are generally not harmful.
- **Kidneys:** Many people with TSC have growths called angiomyolipomas (AMLs) and/or cysts. These usually do not cause problems, though there is a risk of bleeding, usually in late teenage or early adulthood. Around 5% of people with TSC go on to develop many cysts in the kidneys (polycystic kidneys) which can be harmful and on very rare occasions may require dialysis or kidney transplant.

- **Lungs:** Cysts can occasionally occur in the lungs (this is called lymphangiomyomatosis or LAM). These are rare, occurring only in 1-3% of cases.
- **Brain:** There are a number of different types of TSC growth that can occur in the brain. On rare occasions these can continue to grow, and may block the drainage of fluid from the brain (hydrocephalus). This may cause severe headaches, visual problems, muscle weakness and vomiting, and requires urgent medical treatment should it occur.
- **Epilepsy:** This is often a symptom of TSC, and usually develops in childhood. TSC-related epilepsy cannot always be controlled successfully.
- **Learning difficulties:** Because of the growths in the brain and epilepsy, TSC can sometimes affect a child's development and learning. Less than half of people who have TSC have learning difficulties, and if they have not developed by adolescence, it is unlikely that they will develop later.
- **Other TSC-related problems:** Some people with TSC have completely normal intellectual abilities, but find they have problems with specific areas such as memory; information processing and organisation; attention and concentration (ADHD); sleeping; depression and anxiety; and social skills (autistic spectrum disorders).

What treatment and help is available?

Many of the potential symptoms of TSC can be controlled by medical interventions. In particular, there are a broad range of anti-epileptic drugs and other treatments which are sometimes successful in controlling epilepsy. There are also drugs being trialled which may be successful in controlling growths in the lungs and kidneys.

Children affected by TSC may need extra help at school. Every school has a special educational needs coordinator (SENCO) who should be able to help with this. There are also services available to help young people with finding work and education. Contact Connexions in England, or Careers in Wales Scotland or Northern Ireland for further information.

What causes TSC?

TSC is caused by an alteration in a gene. Genes are units of information which tell the body how to work, grow and develop. The genes which may be altered in TSC normally regulate how fast the cells of the body grow. The alteration in TSC causes this regulation to be less effective, so some cells grow faster than they should.

The gene alteration which causes TSC can be passed down through families. However, the alteration can also happen spontaneously. In more than 70% of cases of TSC the gene alteration is new (*de novo*).