

schools, as well as Social Services and the Voluntary Sector are all frequently involved.

For children with autistic spectrum disorders, Local Educational Authorities need to provide a highly structured environment and teaching programme. Ideally parents/carers and schools should work together to provide a united approach to dealing with autistic behaviours. Proper assessment of the child with a learning disorder and behavioural difficulties is vital.

Home teachers, play groups, toy libraries, speech therapy and physiotherapy can all help children achieve their full potential. Social workers, occupational therapists, health visitors and community nurses can all give advice and information and the Tuberous Sclerosis Association can also be a source of help, advice and encouragement.

### **Could any other existing members of my family be affected? Will any further children that I might have be affected?**

Once someone has been diagnosed as having tuberous sclerosis it is important that the immediate family is checked out to determine whether or not this is the first family member with TS. In about 70% of cases TS has come as "a bolt from the blue", with no-one else in the family as yet affected. However, people can have TS without any obvious symptoms and it is important for parents and siblings to find out whether they also carry the TS gene, since anyone with this faulty gene has a 1 in 2 chance of passing it on to any child they may have.

### **Is there a blood test for TS? And is there an antenatal test for TS?**

There is good news on this front! Research funded by the TSA has resulted in blood tests being developed. Not everyone with TS or a family history of TS needs a gene test, but the tests can help when there is concern or doubt about the genetic status of

a patient, or a member of a patient's family, or when pre-natal tests might be considered in a future pregnancy. Families considering genetic tests should ask their GP or hospital specialist for referral to their local clinical genetics service which will then arrange for blood samples to be taken. At the moment it is usually not possible to offer pre-natal diagnosis for TS unless the exact mutation for that family has been identified in advance. This means testing the blood of an affected family member. Once this has been done, samples can be taken from other family members for comparison. Unfortunately, in some cases the genetic damage (the so called "mutation") still cannot be found. An unborn baby can be tested for TS (by chorionic villus sampling -cvs - at 12 weeks of pregnancy), providing the mutation of the affected parent has already been identified. Further information can be obtained from the TSA.

### **What can the Tuberous Sclerosis Association offer me and my family?**

Most people are shocked and devastated when they learn that a member of their family has TS. Rarely have they heard of the condition before and they feel totally isolated. The Tuberous Sclerosis Association (TSA) was set up by parents and interested doctors to support people with TS and their families, to increase knowledge about the condition, and to encourage research into the causes and management of the disorder. Still largely run by parents with a personal experience of TS, the TSA recognises the need for families to have access to the latest accurate information about TS and has developed a wide range of literature and educational material (such as videos and a CD-ROM) for both families and health professionals. The TSA also welcomes contact with people with TS as well as their families and the professionals working with them.

The TSA offer support in a number of other ways, including visiting and telephone support, offering advice and information, and putting families in touch with each other.

The TSA employs TS Specialist Advisers to advise on problems relating to TS, visiting as necessary and liaising with the various agencies as appropriate.

The TSA also organises regional and national meetings and conferences and operates a Benevolent Fund. It has been instrumental in setting up a number of specialist Clinics around the country.

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Further information on TSC and the work of the TSA can be obtained from: Mrs. Diane Sanson, Head of Administration, PO Box 12979, Bant Green, Birmingham, B45 5AN. Tel/Fax: 0121 445 6970  
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**Tuberous Sclerosis Association** 

# Tuberous Sclerosis



[www.tuberous-sclerosis.org](http://www.tuberous-sclerosis.org)

This leaflet attempts to answer some of the questions which are often asked by individuals who are newly diagnosed as having tuberous sclerosis or by their parents, other relatives or carers.

Tuberous sclerosis (TS), also known as tuberous sclerosis complex (TSC) due to the complexity of the condition, is difficult to explain because it affects individuals in many different ways and with varying degrees of severity. Some children and adults with TS remain largely unaffected and go through life free from symptoms, whilst others who are less fortunate experience one or more of a range of symptoms.

Tuberous sclerosis is a genetic disorder with about one third of cases having been inherited from a parent. There is a 50% risk of someone with TS passing it on to any child they may have, but only approximately one third of cases are thought to run in families, with the remainder being sporadic cases.

### **What is Tuberous Sclerosis?**

Tuberous sclerosis derives its name from the tuber-like growths on the brain which calcify with age and become hard or sclerotic. These lesions show up as small white patches on a CT-brain scan in most patients with TS. If they have not yet calcified (perhaps in a very young baby) they may not be seen on a CT-scan, but will be seen on MR images

Abnormal TS growths can affect almost any other organ of the body (including the skin, eyes, heart, kidneys and lungs) but they may cause little in the way of problems. Doctors find them helpful, though, in confirming the diagnosis.

### **How is it diagnosed?**

The condition can be diagnosed any time from infancy to adulthood, depending upon the nature and severity of the symptoms developed by the individual patient. There are a number of different signs of tuberous sclerosis and whilst not all are necessary to make the diagnosis, a combination of some of these signs are necessary. They include:

**Skin:** The earliest sign may be white skin patches (depigmented patches), especially on the limbs and body, which sometimes can be seen from birth. They do not cause any problems and often disappear later in life. In addition, red or brown 'birthmarks' are sometimes found on the face, or a waxy looking reddish lesion found on the forehead. As a child grows older other skin signs may develop, including a characteristic facial rash (facial angiofibroma) across the nose and cheeks in a butterfly distribution. This starts as small red dots, but later these spots can become small bumps and the redness may fade. During adolescence or later, small fibromas or nodules of skin may form around finger or toe nails. Another skin sign, a shagreen patch, may be found on the lower back. It is flesh-coloured and has the appearance of orange peel.

**Heart:** Cardiac rhabdomyomas (benign heart tumours) are another early sign of TS. They may develop in the unborn baby and be picked up during routine antenatal scanning. They rarely cause a problem and frequently regress or even disappear altogether after the child is born.

**Epilepsy:** Seizures occur in approximately 70% of people with tuberous sclerosis. They may start at any age, but frequently begin in childhood, often during the first year. They often start as infantile spasms (salaam fits) for which prompt treatment is required. As the child grows older, the fits may change and sometimes they cease altogether. It is likely that the cortical tubers in TS are the cause of the seizures.

**Developmental Delay:** Whilst 40-50% of people with TS have normal intelligence, the remainder have learning and developmental problems ranging from mild areas of difficulty to 25% with severe disabilities. Early intervention is recommended.

**Behaviour:** About 25% of people with TS are autistic and another 25% show aspects of autistic

spectrum disorder, including higher functioning autism or Asperger's Syndrome. Attention deficit and hyperactivity are often found in children, and anxiety, paranoia and depression are commoner in adults. Sleep disturbance can also be a problem and is usually associated with epilepsy.

**Kidneys:** Very occasionally, a baby presents with polycystic kidneys which are picked up antenatally or shortly after birth, and further investigation leads to a diagnosis of TS. About 70-80% of people with TS develop some other abnormality in their kidneys, but frequently these don't cause any symptoms, especially during childhood.

**Lungs:** Only very rarely will someone with tuberous sclerosis present with symptoms of lung disease.

### **How common is it?**

Tuberous sclerosis is more common than generally recognised. This is partly because there are people with the condition who remain undiagnosed, usually because they are symptom free. It is thought to affect around 1 in 7,000 of the population, meaning that there are around 8,000 affected people in this country alone.

### **What is the outlook?**

The effects of TS are very variable and the outlook depends upon the extent of organ involvement and appropriate monitoring and care. About 50% of people with TS are intellectually unimpaired and lead normal lives, with the remainder having learning disabilities to a greater or lesser extent. Contrary to what was believed for many years, the prognosis for patients with TS is very good. The life expectancy for the majority of them is normal, even for those with severe learning disabilities and epilepsy.

### **What is the treatment? Is there a cure?**

Unfortunately there is no cure for TS but there is treatment for a number of its symptoms and patients should be monitored regularly so that

symptoms can be picked up and treated appropriately.

**Epilepsy:** It is important to get expert advice on anti-epileptic drugs for those with epilepsy so that the best possible control can be achieved without over-sedating the patient.

**Skin:** There is variety of treatment possible (including laser) for the facial rash and the advice of a dermatologist should be sought if the rash causes the patient concern. Referral can also be made to a cosmetic service run by the Red Cross, a service which is simple to use and can make an enormous difference.

**Kidneys:** There are a range of possible treatments for the kidney problems and advice should be sought from one of the specialist TS clinics. Contact the TSA for details.

**Heart:** Medical intervention with drugs is occasionally necessary to regulate an erratic heartbeat. Even more rarely is surgical intervention necessary for obstruction to blood flow due to a rhabdomyoma.

**Developmental Delay:** Many people with TS develop normally, but some children begin to show developmental delay when their seizures start and as they grow older it may become clear that they are not keeping up with other children of their age in certain areas of development. They should be properly assessed and appropriate management offered. Speech and communication are particular areas which may need help as early as possible.

**Behaviour:** Behaviour problems and are best tackled early by a co-ordinated multi-disciplinary effort. A bewildering array of professional services including Community and Hospital Paediatrics, Learning Disability Services, Child Psychiatry, Educational and Clinical Psychology, Speech Therapy, Health Visitors, Special Schools and Special Educational Services within main stream