

may find it helpful. There are a small number of specialist TSC clinics in the country and also a number of doctors with a particular interest in the condition. Your doctor can refer you to one of these specialists or seek their advice about particular problems. Visit the website www.tuberous-sclerosis.org or contact the TSA for more information about specialists and clinics.

Planning a family

TSC is a genetic disorder and an affected person has a 50/50 chance of passing it onto any child they may have. A blood test is available which may find the affected gene in an affected person. This information can be used to test a baby during pregnancy. However the test will only show whether the baby has the altered gene. There is no way of knowing whether a baby with TSC will grow up to be mildly affected like its parent or be more severely affected. A new technique is now available called pre-implantation genetic diagnosis (PGD for short). Couples having this treatment conceive using IVF (in vitro fertilisation also known as test tube babies). The embryos are tested for TSC before being placed in the mother with only unaffected embryos being used. It is a relatively new technique so is not available everywhere and some or all of the costs may need to be paid for privately. Other options may be egg or sperm donation. If you are considering starting a family and would like advice ask your doctor to refer you to your regional genetics centre.

Pregnancy and TSC

Many women with TSC have a normal, healthy pregnancy but there are some potential health complications to be aware of. Women with kidney and lung problems may find that these conditions worsen during pregnancy so it is important to talk to your doctor before starting a family. Some anti-epilepsy drugs (AEDs) can affect the development of the unborn baby and some are not recommended if you plan to breast feed. Talk to your doctor before getting pregnant and s/he can change your AEDs to drugs that are safer for the baby. If your pregnancy is unplanned don't stop taking your AEDs suddenly but talk to your doctor first.

HELP AND SUPPORT

Because the effects of TSC are variable, the amount of support people may need is also variable. Some individuals with TSC don't need any additional help while others do. Some people with TSC may experience difficulties with the following issues:

Education

Many people with TSC have no difficulties at school or college and gain qualifications and jobs, but some find that their TSC affects their learning (see above). Young people with special educational needs and disabilities can get extra help at school and at college. Every school has a special needs co-ordinator (SENCO) who should be able to help. Careers or Connexions services can help young people aged 13 to 19 (up to 25 for those with disabilities) with education and finding work. To find your local service call Connexions (England) 080 800 13 2 19 or visit www.connexions-direct.com, Careers Wales on 0800 100 900 or www.careerswales.com, Careers Scotland on 0845 8502 502 or www.careers-scotland.org.uk, Careers Service N. Ireland on 028 9044 1781 or www.careersserviceni.com

Employment

Some people with TSC may find it difficult to get and keep work. This is sometimes because of problems with epilepsy, poor health or mental health problems. It is unlawful for employers to discriminate against someone because of their disability. If you think you have been discriminated against the Equality and Human Rights Commission can give advice and help you make a complaint, phone 0845 604 then 6610 (England), 8810 (Wales), 5510 (Scotland).

Disability discrimination

Many individuals with TSC will have rights under the Disability Discrimination Act 1995. It makes it unlawful for you to be discriminated against in employment, access to goods, facilities and services, buying and renting property and education. If you think you may have been discriminated against contact the Equality and Human Rights Commission 0845 604 then 6610 (England), 8810 (Wales), 5510 (Scotland).

Travel

Many adults with TSC-related conditions, such as epilepsy, autism or other disabilities, may be eligible to apply for a Disabled Person's Travel Pass, which provides free access to public transport (buses, trains etc). The pass is generally issued to anyone who would be refused a driving licence on medical grounds. To find out if you are eligible, you will need to contact your local council, complete an application form and show how you meet the eligibility criteria.

Insurance

People with medical conditions like TSC sometimes find it difficult to get reasonably priced travel and life insurance and can be refused cover. The TSA is unable to recommend specific insurance providers but may be able to put you in touch with specialist brokers for people with pre-existing medical conditions. Visit www.tuberous-sclerosis.org or contact the TSA for more information.

Financial Support

Disabled people and their carers may be entitled to extra benefits from the government. These include benefits like Disability Living Allowance and Carers Allowance and extra amounts added to Income Support and Tax Credits. Call the Benefit Enquiry Line on 0800 882200 for more information and to get application forms. Your local Citizen's Advice (CAB) or Disability Advice Centre can help you fill them in. There are also trusts and charities that will fund special equipment and holidays. Contact your TSA Specialist Adviser for more information.

Support from the TSA

TSC is a complex condition that is very variable in the way it affects people. It can be difficult for individuals with TSC and their families to understand and come to terms with. The TSA is available to help. TSA Specialist Advisers can provide you with information, advice and support on the telephone, by email and through home visits. They can liaise with the professionals and organisations involved in your care to help you get the best service.

The Outlook Group

The TSA Outlook Group supports those adults with TSC who are relatively mildly affected and interested in contacting and developing friendships with other TSC adults across the UK. Outlook members work in a variety of everyday jobs and professional careers. Some have children themselves. Outlook provides an 'extended sense of community', confidence, support and friendships. It gives people the chance to speak to someone else who understands what living with TSC is all about. The group holds an annual weekend away and many members meet up informally throughout the year. Outlook's motto is: 'I may have TSC but TSC doesn't have me!'

Getting in contact with us

Phone: 0121 445 6970
(Head of Administration who can put you in touch with a Specialist Adviser)
Via the contact page on www.tuberous-sclerosis.org
Email: diane.sanson@tuberous-sclerosis.org
Write to: Tuberous Sclerosis Association,
PO Box 12979, Barnt Green, Birmingham B45 5AN

Tuberous Sclerosis Association

The Tuberous Sclerosis Association is a Company Limited by Guarantee.
Registered in England No. 2900107. Registered Charity No. 1039549.

Tuberous Sclerosis – a guide for people diagnosed when teenagers or adults.



Most people have never heard of Tuberous Sclerosis. Even many doctors know very little about it. This booklet tells you what to expect when you are diagnosed and where you can go to get more information and support.

Tuberous Sclerosis is more properly called 'Tuberous Sclerosis Complex' because it can affect many parts of your body, causing a variety of signs and symptoms. You will see and hear it shortened to both TSC and TS. The most useful abbreviation is TSC since this distinguishes it from Tourette's Syndrome, a different, unrelated condition also referred to as TS.

TSC can be diagnosed at any time, usually depending on what signs and symptoms present. The time of diagnosis can vary from before birth to any time in adulthood. It is caused by an alteration in one of the two genes which regulate cell growth. As a result, extra growth of normal tissue develops in organs of the body (eg. brain, heart, eyes, skin, kidneys, lungs). For some people, these growths may not cause any problems but for others they can result in specific problems in the functioning of one or more of the organs. As a result of the growths in the brain many people with TSC have epilepsy and some have learning and/or behaviour problems. However over half of people with TSC do not have learning disabilities. Most people affected by TSC will live a normal lifespan and any problems related to their condition will be monitored and managed by their doctors.

Diagnosis

A diagnosis of TSC is based on finding a number of different medical features. No single feature is unique to TSC. However some features are seen very frequently in people with TSC (called major features) while others are more common in people with TSC but fairly common in the general population (called minor features). To make a definite diagnosis a doctor needs to find two major features.

If you are suspected of having TSC, doctors will do a series of tests. These may include skin screening under an ultra violet light to look for white patches (see below), a brain scan (CT or MRI) to check for growths in the brain, a scan of the kidneys to check for growths and an EEG (brain wave recording) to check for epilepsy. In most cases the results of these tests will confirm whether or not you have TSC.

Doctors may also arrange tests on other members of your family, if they wish, since TSC is a genetic disorder. This means it can be inherited from a parent or passed onto a child. However, over 70 per cent of people with TSC do not inherit it from their parents but as a result of a new gene alteration.

POSSIBLE FEATURES OF TSC

Skin

An early sign of TSC may be white skin patches (hypomalemic patches) on the limbs and body which can sometimes be seen at birth. They do not cause any problems. A rash on the face (angiofibromas) may start to develop in childhood as small red spots across the nose. It is sometimes more noticeable during teenage years as puberty can make it worse. Other skin signs include raised patches of skin on the forehead (forehead plaques), areas of thickened leathery skin - particularly on the lower back (shagreen patch) and small growths around the fingernails and toenails (ungual fibromas). Different types of treatment are available that can remove or minimise the appearance of skin signs. If you are worried or self-conscious about your appearance ask your doctor to refer you to a dermatologist. Your doctor can also refer you to the local Red Cross Camouflage Service who can advise and supply special camouflage cream.

Kidneys

Many people with TSC have growths in their kidneys; angiomyolipomas (called AMLs for short) and/or cysts. They usually do not cause any problems or symptoms. AMLs may grow very slowly during childhood but if they continue to grow there is a risk of bleeding. If symptoms do occur, it is likely to be in the teenage or early adult years. Once growths have been found in your kidneys it is important that your doctor does regular checks (blood tests, urine tests, scans) to see if any treatment is needed. Treatment is available for AMLs that bleed or are at risk of bleeding. In rare cases a kidney may need to be removed. 5 per cent of people with TSC have polycystic kidneys which can also result in kidney failures. A kidney transplant or kidney dialysis where a machine does the job of the kidney are two treatment options from some people whose kidneys are badly affected. In the future drugs may be available to shrink the AMLs and stop them growing. At time of print (2008) drug trials are currently taking place and the early results are encouraging.

Lungs

Very rarely some women (1-3 percent) have symptoms of a cystic lung disease (called Lymphangiomyomatosis or LAM for short). However recent studies have found that many women with TSC have growths in their lungs that do not cause any problems or symptoms. The drug that is currently being tested on TSC kidney patients is also being tested on LAM patients. It is hoped that it will provide a future treatment.

Brain

People with TSC may have growths in their brain (cortical tubers and subependymal nodules). Some may have both types of growth while others may have none. Very rarely people can have a growth in their brain which continues to grow very slowly and can block the drainage of fluid in the brain (hydrocephalus). This tumour is called a subependymal giant cell tumour or SGCT or SEGA for short. It occurs in less than 5 per cent of people with TSC and usually when they are children or young adults. If it blocks the drainage of fluid from the brain, it may cause any of the following, and requires urgent medical treatment: severe headache, visual problems, muscle weakness and vomiting. Contact your doctor if you are worried.

Epilepsy

Not everyone with TSC has epilepsy. It can start at any age but most people who have TSC and epilepsy start seizures in childhood. It is much less likely that someone will develop epilepsy for the first time in adulthood. There are lots of different types of seizure and over time seizure types may change, become more or less frequent or even stop. It is not always possible to control all the seizures that someone with TSC has. There are many anti-epileptic drugs available and sometimes different drugs and different doses need to be tried to find the ones which work best. For a small number of people non-drug treatments such as surgery, or an electrical stimulator positioned under the skin (vagal nerve stimulator) may be of benefit.

Learning Problems

The presence of TSC growths in the brain and epilepsy can affect a child's development and learning. However over half of people with TSC do not have learning difficulties and older children and adults without learning difficulties will not develop them later.

Some TSC related problems

Some people with TSC who have completely normal intellectual abilities find they have problems with some specific areas.

- **Memory** - many people with TSC have memory-related problems. Difficulties with memory may be related to epilepsy, side effects of medication or underlying brain growths. Stress and anxiety can also affect memory.
- **Information processing and organisational skills** - people with problems in this area may find it difficult to plan tasks and complete things on time or cope with changes in routines. Some of these difficulties are sometimes referred to as 'executive control processing' deficits.

- **Attention and concentration** - ADHD (attention deficit hyperactivity disorder) is more common in people with TSC compared to the general population but even those with out ADHD may find themselves easily distracted, impulsive and fidgety.
- **Sleep problems** - people with TSC may wake more often in the night and experience less efficient sleep.
- **Mood swings** - people with TSC are more likely to experience mood swings including aggression and self harm.
- **Depression and anxiety** - is more common in adults with TSC. This can range from mild symptoms of depression or anxiety to severe symptoms that may require mental health assessment and treatment.
- **Social skills** - there are high rates of autism and Asperger's syndrome in TSC. Some people with TSC find it difficult making and keeping friends, even if they may not have autism or Asperger's.

It is often difficult to get help with these sorts of problems. Contact your TSA Specialist Adviser if your doctor is unable to help you or if you are uncertain about who may be able to help you. It can sometimes be helpful to talk to others with similar experiences. The on-line discussion forum at www.tuberous-sclerosis.org/community/ is an opportunity for people affected by TSC to share experiences.

Other parts of the body

TSC growths may be present in many other organs of the body where they usually don't cause problems. For example growths in the heart are often found in children but get much smaller so that they seem to disappear as they grow. The liver, spleen, pancreas, gastrointestinal system and the bones can also be affected. Growths in the eyes are also common and there can be small growths on the gums and dental pits which cause little holes in teeth enamel. Good dental care is therefore important.

LIVING WITH TSC

Medical care

Adults with TSC will be under the care of different doctors depending on which aspect of TSC affects them – dermatologists for skin treatment, kidney specialists for kidney problems, neurologists for epilepsy or psychiatrists for mental health problems. Children are under the care of a paediatrician who monitors them throughout their childhood and works with other specialists they may need to see from time to time. Regular monitoring is important to identify any potential problems. The Tuberous Sclerosis Association (TSA) has published Clinical Guidelines for the care of patients with TSC. You can download a copy from the website www.tuberous-sclerosis.org or contact the TSA for a copy, your doctor