

A blood test is also available which may find the faulty gene in an affected person. This information can be used in future antenatal testing and to test family members where there is uncertainty.

Medical care

Every child with TSC should have a primary or co-ordinating doctor. This could be a children's doctor (paediatrician), a doctor specialising in genetic problems (geneticist) or in epilepsy (neurologist) or a family doctor (GP). The primary doctor is usually the doctor who confirmed the diagnosis. They will monitor a child throughout their childhood and will work with other specialisms a child may need to see from time to time. There are a few specialist TSC clinics in the country and also a number of doctors with a particular interest in the condition. The primary doctor may refer a child to one of these specialists or seek their advice about particular problems. (Contact the Tuberosus Sclerosis Association (TSA) for more information about specialists and clinics, contact details on back page).

Epilepsy

For many young children the first sign that they may be affected by TSC is epilepsy. This may start as infantile spasms in the first two years of life. These last only a few moments and involve a sudden jerk of the trunk and limbs, followed by a stiffening of the muscles. Sometimes infantile spasms are mistaken for colic. Drugs are now available that are good at controlling infantile spasms. As a child grows other types of seizure may develop. It is not always possible to control all the seizures that a child 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 child. For a small number of children non-drug treatments such as special diets or surgery may have some benefits.

Development delay

The presence of TSC growths in the brain and epilepsy can affect a child's development and learning. The effects vary greatly from one child to another and it is difficult to predict how a child will develop. It is thought that very young children who have difficulty to control epilepsy may have more learning problems than children whose epilepsy is well controlled or develops later.

Children with TSC may be delayed in their speech and language development. They may also have attention problems such as a short attention span, be easily distracted or be hyperactive. Some children with TSC will develop autism. If you are worried about your child's development speak to your health visitor or doctor and they can refer you to a local service that helps children with development problems.

Behaviour

Children with TSC may have behaviour problems. It is difficult to know why they have these problems. It may be to do with the tubers in the brain, epilepsy, communication difficulties or difficulties in understanding the world around them due to learning problems. They may also have difficulties in sleeping. Contact your doctor if you are finding your child's behaviour difficult to manage. Your doctor will want to make sure there isn't a medical problem causing the behaviour and if not can refer your child for specialist help.

Heart

Many children with TSC are born with growths in the heart (cardiac rhabdomyomas). These very rarely cause problems and get smaller as a baby grows. In most cases they will have disappeared after the first year of life.

Kidneys

Many children will have growths in their kidneys (angiomyolipomas or AMLs for short) which usually do not cause any problems or symptoms. They 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 a child's kidneys their doctor will do regular checks (scans, blood tests, urine tests) to see if any treatment is needed.

One in 100 children with TSC have a very rare kidney condition called polycystic kidney disease (PKD). Children who have PKD will have their blood pressure and kidney function checked every year.

Brain

Children with TSC may have growths in their brain (cortical tubers and subependymal nodules). Some children may have both types of growth while others may have none. Very rarely children 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 astrocytoma or SEGA for short. It occurs in less than 5 per cent of people with TSC but needs urgent treatment. It may cause headaches, visual problems, muscle weakness and vomiting. Contact your child's doctor if you are worried.

Skin

A facial rash (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. If your child is worried or self-conscious about their appearance ask their doctor to refer them to a dermatologist. They can also be referred to the local Red Cross Camouflage Service who can advise and supply special camouflage make-up.

General health

Children with TSC should have the standard childhood checks available to all children. Hearing and eye tests are particularly important since hearing or sight problems can make learning problems worse. Some children may also have growths in their eyes although these don't usually cause problems. Eye checks are also useful in spotting early signs of hydrocephalus (see Brain above). Regular dental checks are important. There can be small growths on the gums and dental pits which cause little holes in teeth enamel. Anti-epileptic drugs can also affect the teeth and gums. There is no reason why children with TSC should not have the standard childhood vaccinations however the risk of a febrile convulsion after vaccination means that children with TSC should be monitored carefully and treated with paracetamol to reduce fever should this occur.

Social services

Your local social services department can provide practical assistance in the home, leisure services like play schemes and friendship schemes, home adaptations and help with equipment, babysitting services and respite care to give families a break. As a parent you have a legal right to ask for your child's needs to be assessed and for your own needs as a carer to be assessed. Many families do not like to ask social services for help and are worried about what might happen or what people might think. However most areas have specialist social workers who only work with disabled children and have an understanding of the difficulties families have.

Education

Many children with TSC attend mainstream schools and nurseries, some with extra support. If a school feels it is unable to meet a child's needs it may ask external agencies to help or ask the local education authority to make an assessment of the child's special educational needs - called a multi-disciplinary assessment (MDA). Many people are involved in the assessment including the school, the child's parents, doctors and educational psychologists. A document called a Statement of Special Educational Needs may be issued which sets out how the needs will be met including whether the child needs any extra support and how much. Some children may need their own support worker to work with them all of or some of the time and for a small number of children their needs may be met at a school for children with special educational needs. If there are concerns about a child's development from an early age the local education authority can make this assessment before they start school. Current Government policy is to reduce the number of statements issued and some local education authorities have decided not to issue any statements at all. A meeting should be held every year to review the educational progress of children with statements. Schools should also regularly review the level of support they give to children without statements. Some children may need less support as they grow older while others may need more. Each local education authority has a Parent Partnership Service which is an independent source of advice and support for parents.

Financial Support

Disabled children and their carers are 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. There are also trusts and charities that will fund special equipment and holidays. Contact a TSA Specialist Adviser or your social worker for more information (contact details below).

Voluntary and community organisations and support groups

Local groups can be a great source of local information and support. Many areas have general support groups for families whose children have a variety of special needs. Some may offer services such as babysitting, transport, leisure and family activities. Some areas have local groups for children with specific difficulties such as epilepsy, autism or communication difficulties.

Support from the TSA

Not knowing what the future holds can be very difficult for parents and other family members. The TSA is available to help. TSA Specialist Advisers can provide information, advice and support on the telephone, by email and through home visits. They can liaise with professionals and organisations involved in the care of your child to ensure your child is getting the best service. The TSA also runs regular events where families meet to share activities and experiences and a group of mildly affected adults (the Outlook Group) meets annually and provides mutual support to members throughout the year.

For further information about Tuberous Sclerosis and the work of the TSA contact the Head of Administration who will put you in touch with a Specialist Adviser for your area.

Further information on TS and the work of the TSA can be obtained from: Diane Sanson, Head of Administration, PO Box 12979, Barrt Green, Birmingham, B45 5AN.
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Tuberous Sclerosis Association

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The Child with Tuberous Sclerosis

