

Health Professionals

This guide is written for staff working in the healthcare professions who may work with people living with Tuberous sclerosis complex (TSC) now or in the future. Your knowledge, support and care could significantly improve health outcomes for people living with TSC.

About TSC

TSC is a multisystem genetic disorder with a range of physical features that require lifelong evaluation, surveillance and management. TSC can cause benign tumours to grow in one or more organs of the body. It can affect every organ system, with some more common during infancy and childhood and others more likely to affect individuals as adults. In the UK there are up to 10,000 people living with TSC.

Diagnosis is based on a careful physical examination, in combination with possible scans of the brain, lungs, liver, heart and kidneys. Genetic testing can be used to diagnose and/or confirm a diagnosis of TSC.

Getting the right care and management is crucial for improving the quality of life of people living with TSC and those who care for them. An important part of this care is the coordination of medical professionals from a range of specialities. This is particularly crucial during the transition from paediatric to adult health care.

Some of the main features include:

- **Epilepsy is the most common neurological feature of TSC, occurring in 85% of individuals. Epilepsy is often difficult to control and at least 50% of people living with TSC develop refractory epilepsy (drug resistant epilepsy).^[1]**
- **About 35% of children living with TSC will have experienced infantile spasms.^[1]**
- **Kidney tumours (angiomyolipomas or AMLs) are common and may not cause any problems until adulthood. There is also an increased risk of cysts in one or both kidneys.^[1]**
- **TSC can affect the skin and lead to several conditions including reddish bumps or a rash on the face (angiofibromas) and small growths around the fingernails and toes (ungual fibromas).^[1]**
- **Lymphangiomyomatosis (LAM) is a lung complication of TSC which is most common in women between puberty and menopause.^[1]**
- **Heart tumours (cardiac rhabdomyomas) occur in about 50% of people living with TSC. These generally become smaller over time and only a few people will have long term problems.^[1]**

- People with TSC may have an overgrowth of the gums (fibromas) and dental pits (weakened enamel of the teeth).^[1]
- Cysts and AMLs may occur in other organs such as the pancreas, ovaries or liver.^[1]

International guidelines on the treatment and care of people living with TSC are available here - <https://bit.ly/2xTPQJc>. A set of UK guidelines is due to be published winter 2018.

TSC Associated Neuro-Psychiatric Disorders (TAND)

People living with TSC also have a range of Neuro-Psychiatric issues. Around 90% will have some of these difficulties during their lifetime. These often represent the greatest challenge of the condition and include:

- Behavioural - examples include repetitive or ritualistic behaviour, poor eye contact, shyness, hyperactivity, restlessness and impulsivity.
- Psychiatric - the most common psychiatric diagnoses in TSC are autism, ADHD, anxiety and depression.
- Intellectual – there is a wide range of intellectual ability within TSC ranging from high to low.
- Academic – there is a wide range of academic ability in TSC, and even those without intellectual impairment, may still need support with their learning needs.
- Neuropsychological – this covers brain skills, such as communication and language, memory, attention, planning and visuospatial skills.
- Psychosocial – covers quality of life elements such as self-esteem, relationship difficulties and family stress.

Monitoring any behavioural or psychological change is important as this may also indicate a change in the physical health of a child or adult living with TSC.

A TSC TAND Checklist has been developed to help families and people living with TSC have a conversation with their clinicians to identify difficulties early and to prioritise treatment. Items listed below can be found on the Tuberous Sclerosis Association (TSA) website- www.tuberous-sclerosis.org.

- The TAND checklist can be found here - <https://bit.ly/2ObP3wn>.
- Guidelines for the Assessment of Cognitive and Behavioural issues in TSC can be found here - <https://bit.ly/2Faz6yE>.
- The Neuropsychiatry of Tuberous Sclerosis - A presentation on YouTube by Prof Petrus de Vries can be found here - <https://bit.ly/1xhVTSX>.

Treatment

- TSC is a lifelong condition that requires help and support from a range of professionals. Medical notes are often lengthy and complex.
- There are range of medical treatments that will be targeted towards specific elements of the condition. These include anti-epileptic drugs or mTOR inhibitors (which might be delivered as oral or topical preparations). Some people may benefit from surgical interventions.
- People living with TSC may have frequent medical appointments often at different hospitals or clinics. This can feel overwhelming and help with managing appointments can make a big difference.
- There are multidisciplinary TSC Clinics throughout the UK, contact the TSA for details of your nearest TSC Clinic.
- Research is ongoing to find new treatments and to make them available to all who need them.

Support for Families, Parents and Carers

The complexity of TSC is often difficult for families, parents and carers to understand. It is helpful if appointments can be extended or separate appointments made available for carers or family members. The wellbeing of the carer is important, and referrals need to be made to carers organisations if appropriate.

Further Learning – Free BMJ Module

The TSA has worked with the British Medical Journal (BMJ) to develop a module on TSC. It is designed to help health professionals recognise TSC and offer more information about the treatments currently available. The module is available for anyone to access and is free of charge.

Users who successfully complete the module will receive one Continuing Professional Development (CPD) point.

Register at <https://learning.bmj.com/learning/home.html> to access this module.

Support from the Tuberos Sclerosis Association (TSA)

- The TSA is the only UK charity dedicated to supporting people affected by TSC.
- We have a team of advisers around the UK offering help and support to people living with and those affected by TSC- see www.tuberos-sclerosis.org for more details.
- Support is available to professionals who are working with people who are living with TSC.

For more information about the TSA and its work go to www.tuberos-sclerosis.org or contact admin@tuberos-sclerosis.org or 0207 922 7731 (main switchboard)

Date of Information: September 2018; revise by September 2020

We can provide this information in other formats such as large print or audio and in other languages

Disclaimer

We have made every effort to ensure that information in this publication is correct at the time of going to print. We do not accept liability for any errors or omissions and policy and practice may change.

Reference Website:

1. Tuberos Sclerosis Alliance (2018) *Signs and Symptoms of TSC*, 6 Sept 2018 [<https://www.tsalliance.org/about-tsc/signs-and-symptoms-of-tsc/>]

Supported by a grant from Novartis Pharmaceuticals UK Ltd.

Tuberos Sclerosis Association is a Company Limited by Guarantee registered in England and Wales No.2900107
English Registered Charity Number 1039549, Scottish Registered Charity Number SC042780
Registered office: CAN Mezzanine, 32-36 Loman Street, London SE1 0EH