Understanding TSC

This booklet has been especially developed for parents or carers of children who have recently been diagnosed with Tuberous Sclerosis Complex (TSC). It has been designed to help you understand the condition and assist you with your child's treatment pathway.

This booklet has be developed and funded by Novartis and endorsed by the Tuberous Sclerosis Association (TSA).

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Continued research and improved medical care now allows patients with TSC to live healthier lives, with a normal life expectancy in most cases.

It is really important that potential features of TSC are detected quickly and managed correctly. It is likely that a paediatrician will be overseeing your child's care (your 'primary clinician'), but correctly managing all potential manifestations of TSC can involve seeing many other health professionals at different times.

This guide is designed to help you to understand what the different features of TSC are, when they are most likely to develop and who the relevant health specialists are that you should be put in touch with who can help manage your child's specific TSC manifestations and prevent further complications. This is particularly relevant as services vary across the UK.

What is TSC?

Each month ten babies are born in the UK with Tuberous Sclerosis Complex (TSC).[1,2,3,4] TSC is a genetic condition that can lead to growths in different organs of the body, with the most commonly affected being the brain, skin, kidney and eye.[4] The severity of TSC is very varied. Some people are mildly affected and experience few problems whereas others are more severely affected.[5]

Different aspects of TSC affect an individual's life at different times. Some people will be diagnosed at a very early age (sometimes before they are born) whilst others may not be diagnosed until later childhood, adolescence or even adulthood. It is important to understand that the condition varies significantly from person to person and that most individuals will not develop all of the features described.[4,5]

There is no single feature that is absolutely specific to TSC, however it is normally detected following the finding of heart growths prenatally or in new-borns, the onset of infantile spasms in babies, a diagnosis of autism in children, the development of skin problems in adolescents or the onset of kidney problems in adults.

Symptoms (At Birth)

HEART

Heart tumours (cardiac rhabdomyomas) occur in approximately 50% of babies born with TSC. These tumours, like all tumours associated with TSC, are non-cancerous (benign). These tumours are at their largest at the time of the baby's birth and get smaller over time, eventually disappearing and therefore rarely causing medical complications.[6]

WHAT TO LOOK OUT FOR

The heart tumours are predominately detected during the 20 week scan or at birth. Monitoring of the heart using an echocardiogram (ECCO) and/ or an electrocardiogram (ECG) is recommended when there are symptoms of an irregular heartbeat (arrhythmia).[7]

WHO TO SEE

Paediatric cardiologist at birth or referral from the primary clinician

Symptoms (0-2 years)

EPILEPSY

Epilepsy is the most common neurological feature of TSC and is caused by abnormal areas of nerve cells in the brain.[8] Approximately 40% of children with TSC, usually under the age of two, will have a type of epilepsy called infantile spasms. It is crucial that parents and carers are aware of these symptoms as they are often mistaken for other conditions such as colic or heartburn. In order to reduce the potential long-term complications of epilepsy, including behavioural and developmental disabilities, it is important that it is promptly diagnosed and treated.

WHAT TO LOOK OUT FOR

At first seizures may be as subtle as a gentle head nod or thrust of the chin. Over time these tend to become more obvious jerking movements, which are usually brief and occur several times in a row. A lack of awareness and alertness is also a key sign of infantile spasms. Most commonly these happen either first thing in the morning or after waking up from a nap. An electroencephalography (EEG) is necessary for patients having seizures to try to identify the specific seizure disorder. There are a number of medicines which can be used to control seizures occurring in patients with TSC, however it is important to balance potential side effects with the potential positive effects of any medicine and this should be discussed with the healthcare professional responsible for this part of your child's care.[9]

WHO TO SEE

Your primary clinician or local paediatric service. You may also be referred to see an epilepsy specialist and have regular support from a specialist epilepsy nurse.

Symptoms (2-5 years)

SKIN

Most patients with TSC have changes to their skin. Some TSC skin features may appear at birth or in childhood and therefore are often used to diagnose the condition in young patients.[13]

WHAT TO LOOK OUT FOR

The skin lesions can include light coloured spots or bumps on the skin in several different locations. Yellowish-brown or skin coloured plaques of variable size and shape usually located on the forehead or scalp frequently occur at any age and can be seen at birth or early infancy. Leaf-shaped or polygonal white spots are usually present at birth or infancy and are common on the trunk or buttocks. In the second to fifth year of life, it is common for red to pink bumps with a smooth surface to be found on the chin and the cheek area near to the nose and centre of the face, becoming more prominent with age.[13]

Currently available treatment methods vary and are only needed when the skin lesions are obvious or distressing.[9]

The more common interventions include surgery, dermabrasion, peeling and surgery. Over the last decade lasers have become a popular treatment option and topical creams may also be available.[13]

WHO TO SEE

Your primary clinician or dermatologist.

Symptoms (2-5 years)

DEVELOPMENTAL AND BEHAVIOURAL DIFFICULTIES

There are a number of developmental and behavioural difficulties associated with TSC.[10] Developmental delay is the term used when a young child is slower to reach development milestones than other children. Delay may occur in the way a child moves, communicates, thinks and learns, or behaves with others. If the delay is ongoing, the child may be diagnosed with a developmental or behavioural disorder including autism spectrum disorder or attention deficit hyperactivity disorder (ADHD). The likelihood these issues arising are greatest in children who present with infantile spasms.[11]

WHAT TO LOOK OUT FOR

Some examples of difficult behaviours that are common in people with TSC are unusual social behaviours, temper tantrums, preoccupations, poor eye contact, special interests and speech and language delay. The treatment and management of the disturbances requires well co-ordinated care from a range of different services. If developmental delay or psychological problems are suspected, it is important for the person with TSC to be assessed at key developmental milestones in order for problems to be identified early and to gain access to specialist social services.[11]

WHO TO SEE

Your primary clinician or local paediatric service can recommend other healthcare professionals that are appropriate. This can include specialists that help children with developmental and behavioural problems such as the local Child and Adolescent Mental Health Services (CAMHS). The Tuberous Sclerosis Association has also produced a helpful guidelines document for the assessment of cognitive and behavioural issues in TSC, which can be found at http://www.tuberous-sclerosis.org/Info-and-support-resources.html[12]

Other symptoms to be aware of

KIDNEYS

Although many kidney abnormalities in TSC cause no problems, some can have an effect on the proper functioning of the kidney. The most common kidney problems in people with TSC are angiomyolipomas, which are benign tumours. These occur in up to 80% of people with TSC.[14] Angiomyolipomas begin to grow in childhood but may not cause a problem until early adulthood. Once identified angiomyolipomas require on going monitoring.

WHO TO SEE

Your primary clinician who can refer you to a kidney specialist for monitoring or testing.

EYES

Nearly 50% of individuals with TSC have problems with their eyes caused by development of benign tumours. However, growth and change is rare so usually no treatment is required.[9] Ophthalmologists are able to check for these growths so it is important to have regular check ups.

WHO TO SEE

Optometrist/ophthalmologist.

Other symptoms to be aware of

TEETH AND MOUTH

Individuals with TSC may have an overgrowth of the gums (fibromas) and dental pits (weakened enamel of the teeth). As a result, the teeth and gum tissue are slightly different and require a little more care to keep healthy so regular check ups at the dentist is crucial.[9]

WHO TO SEE

Dentist.

Where can I get support?

The TSA (Tuberous Sclerosis Association) is a UK patient organisation, which was formed in 1977 by a group of parents, affected individuals and healthcare professionals interested in promoting greater understanding of the condition and providing mutual support for affected families.

http://www.tuberous-sclerosis.org

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Appointment Diary

Date and Time	Why has the appointment been made?	Name of Dr/ Nurse/Specialist	Department	Referred from

Useful Contact Details

Primary Clinician Name	Contact details

Specialist / Local Centres

Address	Contact Number	Name of Dr/Nurse/Specialist	Specialty/area

Appointment Diary

Date and Time	Why has the appointment been made?	Name of Dr/ Nurse/Specialist	Department	Referred from

Test Results

Date	Test	Results	Next Steps

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Test Results

Date	Test	Results	Next Steps

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