

# Scan Facts

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Fact-Sheet No 1 of the Tuberous Sclerosis Association

## **EPILEPSY IN TUBEROUS SCLEROSIS**

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# EPILEPSY

Epilepsy is one of the most common features of Tuberous Sclerosis. It is often the problem which leads to the diagnosis of Tuberous Sclerosis being made. Epilepsy occurs in most but not all people with Tuberous Sclerosis.

## **What is epilepsy?**

Epilepsy is the tendency to have recurrent, unprovoked epileptic seizures. Unprovoked means that the epileptic seizures occur out of the blue by themselves and have not been provoked by something such as a low blood sugar, flashing lights or an acute brain injury.

There are many different types of epileptic seizure and different patterns of epilepsy.

The thing that is common to all epileptic seizures is that they are caused by a sudden and excessive burst of the electrical activity of the nerve cells of the brain. The brain is made up of many billions of nerve cells (neurons). These nerve cells send their messages to each other and to other parts of the body by small electrical signals. If the nerve cells are behaving in an abnormal way, they may produce a sudden and very large electrical discharge (rather like a flash of lightning). If this abnormal electrical activity spreads to a large enough group of the nerve cells then it will result in an epileptic seizure. In theory epileptic seizures can arise from any group of nerve cells within the brain and therefore there are many different types of epileptic seizures that can occur. If the epileptic seizure comes from a group of nerve cells on one side of the brain then this is called a partial seizure. If both sides of the brain are involved from the beginning of the seizure then this is called a generalised seizure.

## **Why does epilepsy occur in Tuberous Sclerosis?**

In Tuberous Sclerosis the brain has developed abnormally before birth. The “tubers” from which TS gets its name are abnormalities of the way in which the outside part of the brain (cortex) has developed. The nerve cells in these areas are very abnormal and behave in an abnormal way. It is from these abnormal areas that the epileptic seizures originate. As most people with TS have many tubers, the epileptic seizures may start from several different places in the brain.

A simple way to classify the different epileptic seizure types is into partial seizures (seizures arising from one part of the brain), generalised seizures (seizures starting

from both sides of the brain simultaneously) and partial seizures with secondary generalisation (seizures that start in one part and then spread to involve both sides of the brain). If the seizures are coming from several different places within the brain, this is called multifocal epilepsy. People with Tuberous Sclerosis often have several different types of epileptic seizure. In reality the seizures that occur in Tuberous Sclerosis may show features of both partial seizures and generalised seizure and it is often difficult to be absolutely certain of the type of seizure without complicated investigations.

The type of seizures that a child is having may change with time. It is very common for children who have started with infantile spasms (see below) to later on develop different types of partial and complex partial seizures. They may change again at around the time of puberty. Change in seizure type usually reflects development of the brain. Sometimes, however, seizure types may change after a new treatment is started.

A change in the seizure pattern or type is one of the ways in which the brain tumours associated with tuberous sclerosis can present. If the seizure pattern changes therefore, this should be discussed with a doctor and consideration given to repeating a brain scan.

The following is a description of the main types of epileptic seizures that occur in Tuberous Sclerosis.

### **Infantile spasms**

For many children with TS the first seizures that they are recognised to have are infantile spasms. With hindsight it is often recognised that children with infantile spasms due to TS have been having some form of seizure before the spasms develop. These are often quite subtle partial seizures which may involve stiffening of one limb, movement of the eyes to one side, jerking of one limb or the face, head turning, grimacing or other subtle movements. These may not be recognised as epileptic initially. *Infantile spasms* involve a sudden jerk of the trunk or limbs followed by a brief stiffening of the muscles involved. The whole movement may last from 1 second to 5 seconds. Typically these movements occur in clusters where one spasm is followed by another after a variable length of time. A cluster may last from 5 to 20 minutes and many spasms may occur in a cluster. Spasms may cause flexion of the trunk and bending at the middle, the arms are often elevated and thrown forward and the legs may

also extend or flex. In TS these movements are often asymmetric and one arm may bend and the other stiffen or the head may be turned to one side. During the spasms the child may cry and often becomes increasingly irritable during the cluster. Infantile spasms in TS most commonly occur at around 4 months of age and rarely after the age of 11 months or before 3 months. Exceptionally spasms can begin in the neonatal period. Spasms may occur after the age of 2, in which case they are called epileptic spasms. These are not uncommon in TS and are usually a form of partial seizure in which one side of the body or one limb will suddenly jerk and go stiff. These seizures also tend to occur in clusters.

## **Partial Seizures**

There are many different types of partial seizures. What is common to them all is that they are seizures starting from one part or side of the brain. Partial seizures which do not cause alteration in consciousness are called *simple* partial seizures. Partial seizures with alteration in consciousness are called *complex* partial seizures.

### ***Simple partial seizures***

This type of seizure can cause either abnormal movements or abnormal feelings. The most common type of partial seizure involves abnormal movements of one side of the body. This might be jerking movements of the side of the face, an arm or the legs. Sometimes there is only stiffening of the limb (tonic seizure). Sometimes there is a combination of stiffening followed by jerking (tonic clonic). The movements may be confined to the face with turning of the head or eyes to one side (the so-called adersive seizure). Sometimes seizures will start in one place and spread to another, for example jerking of the face spreading to involve the arm and then the leg.

In some partial seizures there is only an abnormal sensation felt in one part of the body or more vaguely in the head. These can include tingling feelings, numbness and occasionally other sensations. They often occur together with abnormal movements.

### ***Complex partial seizures***

This type of seizure is common in Tuberous Sclerosis and can result in a wide range of different behaviours, some of which can be quite unusual. Common to all complex partial seizures is an alteration of consciousness. Usually this is

not complete and can range from a slight dreaminess and alteration in normal behaviour to complete unresponsiveness and lack of awareness. Usually people have no memory that the seizure has taken place.

Complex partial seizures typically begin with an *aura*. This may be simply a funny feeling which may be in the head, in the throat, in the abdomen or may be more complex involving hallucinations such as funny smells, funny tastes, a sense of familiarity or a fear. Young children may suddenly stop what they are doing and appear to be frightened or preoccupied. The aura may last only a few seconds or sometimes is much longer. This is then followed by altered awareness and frequently by automatic repetitive behaviour or *automatisms*. Typical automatisms include lip smacking, sniffing, swallowing, fumbling with clothes or fidgeting. This may represent the whole seizure which after a few minutes will stop and is then usually followed by tiredness or falling asleep. However, not infrequently this stage of the seizure will then evolve either into a partial motor seizure with shaking or stiffening of one side of the body or possibly into a generalised seizure (see below). It is very common in this type of seizure for there to be changes in colour such as flushing or pallor, sweating, a fast heart rate, sometimes widening of the pupils of the eye and changes in the breathing pattern. These are called autonomic changes and can occur in any type of epileptic seizure.

Complex partial seizures may be very subtle and especially in young children may not immediately be recognised epileptic.

### **Generalised tonic clonic seizures**

This is probably the best known type of epileptic seizure and is what people used to refer to as *grand mal*. In these seizures there is sudden loss of consciousness from the beginning of the attack. This can result in a fall to the ground. There is then stiffening of the arms and the legs, usually with the arms and legs extended but sometimes bent. During this time the teeth are often tightly clenched and the eyes often are turned upwards. The lips at this point will often go blue and the breathing may stop or be very shallow. Sometimes at the start of the seizure there is a loud cry or funny noise.

This is the *tonic* phase of the seizure. After some time the limbs begin to jerk in a rhythmic fashion. There may also be jerking of the sides of the mouth and grimacing. Gradually the jerks become slower until the seizure stops.

During a generalised tonic clonic seizure there is often excessive production of saliva and secretions which results in dribbling and drooling.

After a generalised tonic clonic seizure there is usually tiredness, confusion, sometimes a headache and it is often followed by a deep sleep. Generalised tonic clonic seizures are often preceded by a partial seizure. This is called secondary generalisation. Most tonic clonic seizures occurring in TS are of this sort.

### **Other seizure types**

Myoclonic seizures are sudden electric shock like contractions of muscles which result in a single sudden jerk of the affected muscles. They can be partial or generalised and can cause a jerk of the limbs, the face or the trunk which may result in falling to the ground. In TS pure tonic (stiffening) seizures may occur.

*Drop attacks* are seizures in which the child falls or is thrown to the ground. These can be caused by a myoclonic seizure, a tonic (or stiffening) seizure or by an *atonic* seizure where the muscles suddenly relax and the child slumps to the ground. Drop attacks not infrequently result in nasty injuries such as cutting the chin or back of the head.

*Absence seizures* are seizures in which there is alteration or loss of consciousness without any abnormal movements or convulsions. Seizures like this do occur in Tuberous Sclerosis and are really a type of simple or complex partial seizure (see above). Typical childhood absence epilepsy or *petit mal* is **not** a feature of Tuberous Sclerosis.

There are many other abnormal behaviours which can be a result of a seizure. It may sometimes be difficult to determine whether an abnormal behaviour is epileptic or not.

### **Status Epilepticus**

Sometimes epileptic seizures are very prolonged. If a seizure goes on for half an hour or longer or if repeated short seizures occur over a period of half an hour or longer without consciousness being regained this is called status epilepticus. There are as many types of status epilepticus as there are types of epileptic seizure. Status epilepticus is a medical emergency, particularly if the seizure is a generalised tonic clonic seizure. Prolonged seizures such as these can cause death due to breathing problems, lack of oxygen in the blood and inhalation of vomit during a seizure. In addition a prolonged seizure can cause

damage to the brain even if the breathing and oxygen levels in the blood are adequate.

For this reason medical help should always be sought in convulsive status epilepticus.

Prolonged complex partial seizures and other non-convulsive seizures may occur. In these seizures there is prolonged unresponsiveness. The child may appear confused, be unsteady on their feet, drool, fidget and may have minor twitching movements of the limbs, face or the eyes. These episodes can go on for several hours to days. Sometimes non-convulsive status epilepticus may be difficult to diagnose and in these situations an EEG is necessary (see Fact Sheet 28).

### **Does epilepsy increase the risk of death?**

Epilepsy can increase the risk of death in the following ways :

1. *Accidents occurring during a seizure.* This includes drowning in the bath, falls and occasionally other injuries. Prevention of this involves common sense measures.
2. *Death due to status epilepticus* is usually due to breathing difficulties which occur during status epilepticus, especially if there has been inhalation of vomit. A very prolonged seizure (greater than several hours) can subsequently lead to multiple organ failure and death.
3. *Sudden unexpected death in epilepsy.* Rarely someone who has epilepsy will die suddenly and unexpectedly. The exact frequency of this in children is not yet known. However, it is uncommon. The reasons why this happen are not completely clear and there are many theories currently being researched.

### **Investigations**

It is not usually necessary to do any additional investigations to make the diagnosis of epilepsy in Tuberous Sclerosis. Sometimes however, investigations are done for specific reasons.

*Electroencephalogram (EEG)* – This records the brain wave activity. This might be done at the time of diagnosis in order to try and determine whether abnormal movements or behaviours are epileptic or not. Once the diagnosis has been made regular EEG's are not necessary. An EEG is sometimes of value in indicating where in the brain the seizures are coming from.

Sometimes it is useful to do a continuous EEG for a 24 hour period or longer. This may be useful when abnormal behaviours are occurring during sleep and it is uncertain whether these are epileptic or not.

Another type of continuous EEG is a Video EEG where in addition to the EEG recording there is a continuous video made of the child so that as well as seeing the EEG activity, the abnormal movements or behaviours can be looked at at the same time.

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## **Tuberous Sclerosis Association**

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