



## **THE NORTHERN IRELAND TUBEROUS SCLEROSIS (TS) CLINIC**

Tuberous Sclerosis Complex (TSC) is a rare genetic condition which affects the body's major organs including the brain, skin and kidneys. Approximately 85 people in Northern Ireland are known to have TSC. TSC is variable in how it affects the individual, and may appear differently even in members of the same family. Some individuals who have TSC are mildly affected and have very few signs of the condition, but others may be more severely affected and can have epilepsy and/or learning problems. TSC can arise in an individual for the first time in a family, or it can be inherited from parent to child of either sex.

The Northern Ireland TS clinic is one of a small number of clinics around the UK with the aim of providing a local service for families and individuals who have TSC, as well as supporting and working with other health, education and social services staff who are providing services to families with TSC.

### Where and when is the TS clinic held?

The Tuberous Sclerosis clinic is part of the Northern Ireland Regional Genetic Service and takes place in Wing G, outpatients department, Belfast City Hospital. The clinic runs once each month (by appointment only) on the fourth Thursday of the month. In a month where a fifth Thursday falls, two clinics are held. The clinic does not run during July, August or December. Between 13 and 16 clinics are usually held each year. Four appointment slots are available at each clinic, where both adults and children are seen. The TS clinic sees both new patients and

patients who come for review appointments.

### Who runs the clinic?

The TS clinic is under the direction of senior medical staff in the Genetics department and a paediatric (children's) doctor who has international expertise in the management of TS. Clinical Geneticists are medical doctors who specialise in a wide number of medical conditions that run in families. There is also a specialist genetic nurse counsellor who works one half day per week for nine months of the year (days and times may vary). Genetic counsellors have a background in nursing, science or other health related discipline and have undergone further training. Secretarial staff are also involved in managing the TS clinic. They send out appointments and will deal with queries about clinic times and appointments. They are available Monday – Friday 9am – 5pm.

At some TS clinics a doctor in training may be present. This will usually be a doctor who is training to work in a specialist area of medicine such as genetics, paediatrics or dermatology (skin). We appreciate that for families it can feel like there is a lot of hospital staff in the clinic room, but we feel it is important that junior doctors learn more about TSC.

### What services are offered at the TS clinic?

Services at the TS clinic fall into three main areas.

The first is the clinical genetic service, that is, making a diagnosis of TSC in a newly referred patient, genetic testing, and the investigation of other family members and discussing having a family if you are affected with TSC.

The second is ongoing management of TSC in an affected person or family through review appointments.

The third area is out of clinic support from the genetic nurse or counsellor.

### **1. Making a diagnosis of TSC**

A diagnosis of TSC can be made at any age. Below are the most frequent reasons for someone to be referred to the Northern Ireland TS clinic -

- Where the person suspected of having TSC has been admitted to hospital with seizures. In this situation a diagnosis of TSC may already have been made in hospital and the person is then referred to the TS clinic. This is most commonly seen in children, particularly those under one year old, some of whom will be under the care of the paediatric neurology service based in the Royal Belfast Hospital for Sick Children.
- A dermatologist (skin doctor) has referred you because you have skin signs that may or may not be associated with TSC and you need further investigation. This is more commonly seen in an older child or adult.
- There is a history of TSC in your family and you want to know if you or any members of your family are at risk of having the condition, or you are planning a family and you want to talk about this in detail. In this case you can refer yourself to our clinic directly. We may need to confirm medical information in you and possibly other family members (with their permission).

### Before you are seen at the TS clinic

The genetic counsellor may make contact with you before your first clinic appointment to take some background medical information, draw a family tree and discuss the referral. It is important to know if any other family members are affected or have seen the genetics service in the past. Medical information is stored in charts in the genetics department and is held in family files. Confidential information about a family is not shared with other family members without their permission.

### The first clinic appointment

Making a diagnosis of TSC is often not straightforward and it is not always possible to give a definite diagnosis at the first clinic appointment. At this appointment, you or the member of your family who is being investigated for TSC will be examined by the doctor, particularly the skin, eyes and nails. It may be necessary to organise further investigations such as ultrasound scan of the kidneys and a scan of the brain - this can take some weeks to complete. You may be asked back to the clinic several times before a diagnosis can be confirmed. 40 minutes is allocated for each appointment slot, so there is plenty of time for discussion with clinic staff.

It is important to establish if TSC has arisen for the first time in an individual, or if other family members are affected or are at risk having TSC. In the individual who has been diagnosed with TSC (based on examination and investigation results) a blood sample may be taken for genetic testing.

All TSC genetic test samples go to a specialist laboratory service in Cardiff in Wales and it can take 12 – 18 months for the test to be processed. In the majority of people who have signs on or within the body that confirm that they have TSC, a genetic change can be found. In the remainder of people it

means that the laboratory has been unable to find the genetic change, however, there is no change in the diagnosis of TSC. Where a genetic change has been found in an affected individual, genetic testing may be available to other family members.

## **2. Ongoing management of TSC**

Most people who are affected with TSC attend a number of hospital and other appointments. Because the condition is so variable, the way in which TSC affects the daily life and medical needs of the individual is very different. Therefore the frequency of review appointments at the TS clinic varies according to the way the individual is affected. When you attend for review appointment you will usually be told how frequently you need to attend. If you have any concerns outside of appointment times, please contact the clinic (contact number at the end of the page). For people who have TSC and who have epilepsy, this is always managed outside the TS clinic.

Most review appointments take place at yearly or two yearly intervals. Some patients, particularly those who are mildly affected or who see other doctors, don't wish to have regular review appointments at the TS clinic but contact the clinic if they need to.

At the review appointment there is an opportunity to discuss the overall management of TSC and to discuss such things as growth and development in children, education, social support and any new issues or developments which may need referral on to other services. Many carers of adults who have TSC and learning difficulties welcome an opportunity to attend the TS clinic once a year for a general health check and discussion about TSC in their relative. Some of the problems that arise as a result of TSC such as behaviour

difficulties or sleep problems can be very difficult to manage and can be ongoing. Some families find it useful to come to the TS clinic and talk about managing these challenges that they face in day to day living.

At the clinic we can undertake health screening related to TSC, such as arranging routine kidney scans (if this is needed) and measuring blood pressure. We can refer someone with TSC to other specialties if this is necessary, these include dermatology, psychiatry and psychology.

A clinical genetic service is available to review patients at the TS clinic. This may be indicated where someone who has TSC is thinking of starting a family and wishes to discuss the options that are open to them with the clinical geneticist and genetic counsellor. This can include discussion about prenatal genetic diagnosis, where genetic testing has found a genetic change in one of a couple. This technique is arranged through the clinical genetic service and Royal Jubilee Maternity Hospital.

### Support outside clinic times

If you or a family member has been referred to the TS clinic, you have probably been attending a number of hospital clinics recently, or perhaps your child has been admitted to hospital. For many families this is a very stressful and worrying time. You may be unfamiliar with the genetics service and not sure what to expect. You may worry about sharing genetic information with other family members, or asking them for medical information. If a diagnosis of TSC has been made in your child, you may be concerned about what the future holds. You may feel anxious about going through genetic testing, or you may just want to talk about your worries or recent experiences.

The genetic nurse or counsellor can offer support and information to you and

your family outside of clinic times either by phone, or she may be able to meet with you outside of clinic times at Belfast City Hospital. It may be possible to meet with you at one of the other hospitals in Northern Ireland, or at your local health centre (depending on local arrangements). As the genetic nurse counsellor service only operates one half day per week, it may not always be possible to respond quickly to enquiries.

Please leave a message on the voice mail or phone the clinic secretary and we will try to deal with your enquiry as swiftly as we can.

Research is something we feel is important as a way of knowing more about TSC. We may ask if you would be willing to take part in any research projects we are involved in. There is no obligation whatsoever to be involved in research if you do not wish to, and a refusal will in no way affect any treatment you receive at the clinic.

#### What services are NOT offered at the TS clinic?

*Emergencies or serious health concerns:* The TS clinic is a monthly outpatient clinic only and does not offer an emergency or out of hours service. If you or your child is acutely unwell in a way that you think may be related to TSC, you should phone your family doctor or attend a casualty department in the hospital nearest to you.

In particular, if your child is under the care of the paediatric neurology service (based in the Royal Belfast Hospital for Sick Children) and you have a query about worrying symptoms related to TSC or the management of epilepsy, then our advice is always to contact this service directly on Tel 028 90 634237. The TS clinic works closely with the paediatric neurology service in the management of TSC in children.

*Managing epilepsy:* The TS clinic does not manage epilepsy, or change or prescribe epilepsy medication. This is because epilepsy is best managed by a local doctor who is looking after you or your child. In children this is usually a hospital or community paediatrician. In adults epilepsy may be managed by the family doctor, adult neurologist or psychiatrist in someone who has severe learning problems.

#### How can I contact clinic staff?

In the first instance, please call the secretarial staff and leave a message. One of the clinic staff will contact you; this will usually be the genetic counsellor.

Secretarial staff can be contacted on:

Secretary, genetic counsellors-  
Tel 028 90 263034

Secretaries, medical staff -  
028 9026 3872 (Patrick)  
028 90263857 (Shane)

#### Where else can I find help and information about TSC?

At the clinic we always direct families to the UK Tuberous Sclerosis Association as the best source of information. Their telephone number is 0121 445 6970, and website address is [www.tuberous-sclerosis.org](http://www.tuberous-sclerosis.org).

If you wish to speak to someone in Northern Ireland who has been directly affected by TSC, there is a local volunteer representative of the UK Tuberous Sclerosis Association who offers support and friendship to families. Her name is Lynne Murton and she can be contacted on Tel 028 91 459416, email [Ireland@tuberous-sclerosis.org](mailto:Ireland@tuberous-sclerosis.org)