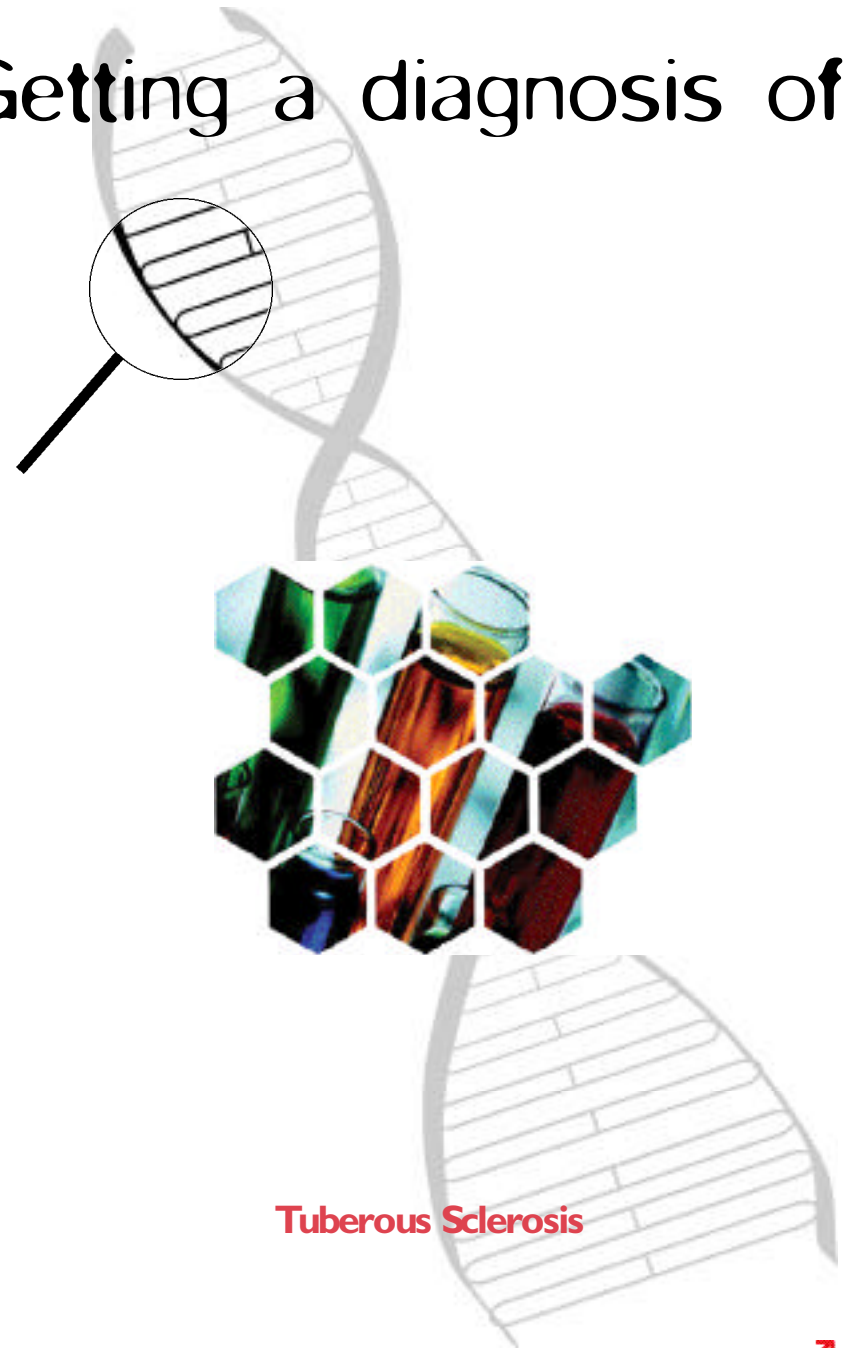


About Tuberous Sclerosis:

- Tuberous Sclerosis (TS) is a genetic condition that causes tuber-like growths to form on the brain, and in other parts of the body.
- TS is very variable, but symptoms can include white skin patches, kidney and lung problems, epilepsy, developmental delay and behavioural problems.
- There is no cure for TS, but some of the symptoms can be treated successfully, especially the epilepsy.
- TS is caused by a genetic change called a mutation. In around a third of cases, the mutation is inherited from an affected parent, but in the remaining two thirds of families, the condition appears 'out of the blue'. People who carry the mutation have a 50 per cent chance (one in two) chance of passing it on to any child they have.
- TS is diagnosed if a combination of the above symptoms are present, and can be confirmed with a brain scan. A genetic test can also confirm the diagnosis, although it is not always possible to identify the exact mutation.

Getting a diagnosis of...



Find out more

To find out more about tuberous sclerosis, and for information and support contact:

Tuberous Sclerosis Association
01993 881238
www.tuberous-sclerosis.org



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Tuberous Sclerosis



Unit 4D , Leroy House ,
436 Essex Road,
London N1 3QP
Tel: (020) 7704 3141
Fax: (020) 7359 1447
Email: mail@gig.org.uk
Website: www.gig.org.uk

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Tuberous Sclerosis

Who should have a genetic test for Tuberous Sclerosis?

Variations in two different genes – called TSC1 and TSC2 – have been identified in the DNA of patients with tuberous sclerosis (TS). DNA variations (or mutations) are uncovered by a process called DNA sequencing, which examines the sequence of letters in the entire DNA code for the genes. This process is time consuming and labour intensive because both genes are large and over 300 different mutations have been found.

All individuals in whom a mutation has been identified in either TSC1 or TSC2 are expected to develop symptoms of the disease. The symptoms of TS vary widely (even amongst affected members of the same family) and it is not possible to accurately predict a person's likely symptoms based on which gene is altered and which particular mutation is detected. Between 20% and 30% of individuals with TS have no mutation identified in either of these genes using current techniques.

If a mutation in one of the TSC genes is identified in a person affected by TS, it may be possible to test future pregnancies by prenatal diagnosis.



Mandy lives in South West London with her husband Philip and their two children, Joe and Ben. Joe, now aged three, was diagnosed with Tuberous Sclerosis (TS) when he was a baby.

Mandy, a GP, first noticed unusual patches of skin that she thought might be caused by TS when Joe was around four months old. At first, her worries were dismissed, until she noticed other symptoms:

'I became aware of the fact he wasn't using his left hand properly- he was able to hold rattles and toys in his right and just didn't seem to be able to do the same with his left, so I became more concerned'.

After seeing a consultant neurologist who arranged for Joe to have a brain scan, Joe was diagnosed with TS when he was six months old. Mandy says she was 'absolutely devastated' by Joe's diagnosis, especially since he was the only person in the family known to be affected. But she said it was helpful being able to put a label to Joe's symptoms:

'It immediately puts you in contact with other groups - in our particular instance there's the TS Association who were fantastically helpful from the outset...as much information as we wanted, and if we wanted to, we could talk to other families going through similar traumas. I think that was the main thing - knowing that we weren't on our own'.

Mandy describes Joe's symptoms, which include developmental delay, as 'fairly severe':

'He has quite severe epilepsy which for six

months was almost impossible to control. It is now controlled but he's on three different medicines morning and night'.

Joe now goes to nursery three mornings a week, where he has a personal helper funded by the local education authority:

'There's no doubt having a definite diagnosis helps when trying to get the benefits to which a family of a child with special needs is entitled - its things like disability living allowance or getting a statement of special educational needs'.

TS is caused by a genetic change, called a mutation. Although Joe's diagnosis was based on his physical symptoms, a genetic test was carried out to confirm that he had TS, and to look for the mutation involved. Meanwhile, Mandy's husband Philip found out that he also had a rare, 'mosaic' form of TS, which means that the condition only affects some parts of his body. The couple were told that as Philip carried the mutation, they had a one in two chance of having another child with TS every time Mandy became pregnant.

'Initially the actual genetic side wasn't that important, it was the clinical side - we didn't need the genetics to get Joe's diagnosis. But as time's gone on, looking at life as a whole and our wish to have another unaffected child, then yes it's been really important that the advances in the genetic understanding of TS have been made. Because Joe's mutation has been identified I can be screened at an early stage of any pregnancy to see whether the fetus has TS.

Mandy became pregnant last year, but opted for a termination after a prenatal test revealed that the fetus was affected by TS. Mandy says this difficult decision was devastating, but she felt that bringing another TS child into the family, who might be more severely affected than Joe, wouldn't be fair on either Ben or Joe.