

Heart

Benign heart tumours (cardiac rhabdomyomas) are an early sign of TS which are seen in about 60% of children. They may be detected before birth on a routine ultrasound scan. They rarely cause any problems, and usually disappear.

Behaviour

Behavioural problems are common. About a quarter of people with TS are autistic and another quarter show some autistic features. Attention deficit disorder and hyperactivity are common in childhood and anxiety, paranoia and depression are more common in adults. Sleep disturbance is also seen more commonly in people with TS.

What treatment is available?

Unfortunately there is no cure for TS. However, many of the different aspects of the disorder can be treated. Treatment will involve a number of different professionals, depending on symptoms. For example, epilepsy should be managed by a neurologist or paediatrician experienced in the management of epilepsy. The skin problems often can be treated and would need referral to a dermatologist (skin specialist). Kidney problems may require the input of a kidney specialist.

What is the outlook?

TS is a variable condition, so that the long term outlook depends on symptoms and severity of the disorder in any individual. About half the people with TS will be intellectually normal and lead normal lives. Others will have a degree of intellectual impairment, but many of these people will have a normal lifespan.

What causes TS?

TS is caused by an alteration in a gene. A gene is a segment of DNA that has a particular purpose. A gene codes for (contains the chemical information necessary for the creation of) a specific enzyme or other protein. Genes determine our personal characteristics such as eye colour and hair colour. We know of two genes that can result in TS, named TSC1 and TSC2. Affected people are found to have an alteration in either TSC1 or TSC2.

About 7 out of 10 people with TS are the first in their family to be affected. This is because the alteration in the gene has arisen in them for the first time. However, when they have children each child has a 50:50 chance of inheriting the altered gene.

The remaining 3 out of 10 people with TS will have inherited it from a parent. Sometimes the parent may be very mildly affected, to the extent that they are not aware that they are affected. Parents of affected children should be carefully examined for signs of TS and, if they are planning further children, gene testing may be performed (see below).

Is there a blood test for TS?

It is now possible to analyse TSC1 and TSC2 to find the gene alteration. The test detects an alteration in most, but not all, affected people. Gene testing is not necessary in all people, but can be offered to those in whom the diagnosis is uncertain or to people who are at risk in a family or to those who might wish to have prenatal diagnosis (a test on a pregnancy). Prenatal diagnosis and family testing can only be offered if the gene alteration has been identified in an affected family member.