

Scan Facts

Fact-Sheet No 22 of the Tuberous Sclerosis Association

CARDIAC INVOLVEMENT IN TUBEROUS SCLEROSIS

by

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Introduction

Cardiac involvement was noted in the first medical report of an infant with tuberous sclerosis (TS) published in 1862. However, the fact that cardiac involvement is common and rarely causes problems was not appreciated until the 1980's, when ultrasound scanning of the heart became freely available.

This fact sheet is written to answer some of the questions commonly asked by TS families when they are told that the heart may be affected by the condition.

How does TS affect the heart?

The commonest finding is one or more rhabdomyomas. These are nodules, usually no more than a centimetre across, which develop within the heart muscle. When these nodules are examined under the microscope, the cells which form them have many similarities to the cells in heart muscle. It seems likely that something goes wrong with the programming of small groups of primitive heart muscle cells while the heart is developing in the first weeks of pregnancy. Instead of developing into mature heart muscle, these cells set off along a disorganised pathway leading to the formation of functionless nodules.

In medical writing, any swelling, lump or nodule can be called a tumour without implying cancer or malignancy. This means that cardiac rhabdomyomas are often referred to as cardiac tumours, but have no connection with any type of cancer.

Other heart problems described in association with TS include a generalised abnormality of the heart muscle, and abnormalities of the heart valves which may be caused by the pressure of a rhabdomyoma within the developing heart. These problems are so rare that the world literature contains only one or two reports and this fact sheet will concentrate on rhabdomyomas and their effects.

How common are rhabdomyomas?

Cardiac ultrasound scans (echocardiograms) demonstrate rhabdomyomas in about 50% of people with TS. Two thirds of these will have multiple nodules throughout the heart muscle.

What problems may be caused by rhabdomyomas?

In the vast majority of cases, rhabdomyomas do not cause any problems at all, and they remain undiagnosed unless cardiac ultrasound is performed. Heart problems due to rhabdomyomas usually develop in early infancy, or even before birth. The problems fall into three groups:

- i) **Obstruction.** Very rarely, one or more large rhabdomyomas block the circulation of blood within the heart and cause heart failure in young infants. If the heart failure is controlled by medication, the problem usually resolves as the baby's heart grows. Surgery to remove the rhabdomyoma is only necessary in exceptional cases.
- ii) **Mass effects.** Extremely rarely, so much of the heart muscle is replaced by rhabdomyomas that the heart cannot support the

circulation independently. Affected babies are usually stillborn, or die within a few hours of birth.

- iii) Disturbance of heart rhythm. Rhabdomyomas may interfere with the normal nerve conduction pathways within the heart, affecting the rate and regularity of the heartbeat. There are no large research studies to demonstrate how many of those with rhabdomyomas have abnormalities of heart rhythm. One study of 11 people with TS found rhythm abnormalities in 7, none of whom had symptoms. Abnormal heart rate may be the first sign of rhabdomyomas in an unborn baby. Where rhythm abnormalities cause symptoms such as palpitations or breathlessness, the problem usually starts in infancy and improves as the child gets older. Symptomatic rhythm abnormalities rarely develop in later life. Troublesome symptoms can usually be controlled by medication.

How are rhabdomyomas diagnosed?

Rhabdomyomas are usually diagnosed by echocardiography. This technique uses ultrasound to produce an image of the heart with its valves and chambers on a screen. The rhabdomyomas show up as dense white areas which are distinct from the adjacent heart muscle.

Echocardiography may be carried out for a number of reasons. Doctors are increasingly aware of rhabdomyomas as a sign of TS and may ask for an echocardiogram to help establish a firm diagnosis. Sometimes, rhabdomyomas are found incidentally, for example, when a child is investigated because of a heart murmur. 10 – 20% of normal children will have heart murmurs and have no signs of heart disease at all. It follows that when a child has a heart murmur and a rhabdomyoma, the two are not usually connected.

What happens to rhabdomyomas?

Rhabdomyomas develop in the unborn baby and steadily increase in size during the second half of pregnancy. They probably reach their maximum size around the time of birth, but then start to regress. Many rhabdomyomas disappear completely: the remainder become smaller and less dense on echocardiography. This means that rhabdomyomas are commoner in children than in adults. Complications due to rhabdomyomas occur almost exclusively in late pregnancy or the first 12 months of life.

Are rhabdomyomas associated with any other signs of TS?

There is no link between the presence of rhabdomyomas and any of the other manifestations of TS.

Unlike all the non-cardiac effects of TS which develop as affected individuals get older, rhabdomyomas become less common with increasing age. They may be the very first sign of TS and can help to confirm the diagnosis before characteristic skin or brain scan changes have appeared.

Can the detection of rhabdomyomas assist genetic counselling?

No one knows whether some families with TS are more likely to develop cardiac rhabdomyomas than others. Where a parent has TS, identification of rhabdomyomas in an unborn baby allows the diagnosis of TS to be made in the second half of pregnancy. Some couples have chosen to terminate a pregnancy in these circumstances.

Can rhabdomyomas be confused with any other cardiac tumours?

There are other kinds of heart tumour which look similar to rhabdomyomas on echocardiography. However, they are extremely rare in young children. When there is a family history of TS, or when the nodules are multiple, it is safe to diagnose rhabdomyomas. A single tumour presenting in a child with no family history or signs of TS causes diagnostic difficulty. It may be necessary to remove or biopsy the tumour in order to find out exactly what it is.

Summary

Most people with rhabdomyomas never know they are there unless they have an echocardiogram.

Rhabdomyomas may be the very first sign of TS in young children, and it is important to examine relatives and follow up the child so that the diagnosis can be confirmed as soon as possible. Similarly, detection of cardiac tumours in infants with seizures or other neurological problems will permit early diagnosis of TS, and echocardiography should be considered in the assessment of these babies.

If you are puzzled by any of the information in this Fact Sheet, your General Practitioner or hospital consultant will be able to explain it further.

References

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This information leaflet is produced for parents by the TSA.
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