TVR Car Club

SPEED CHAMPIONSHIP

Sponsoring:

Marfan Trust

Supporting Research into Marfan Syndrome

www.marfantrust.org

Marfan Trust
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Our Mission

The Marfan Trust is the only Marfan charity in the UK that funds its own laboratory undertaking medical and analytical research so that more is known about Marfan syndrome and its management. The results of our internationally recognised research enable doctors and surgeons to provide better treatment for patients in the short and long term. Over 18,000 people are affected by Marfan syndrome in the UK, with many more anticipated to be living with the condition undiagnosed. Our important work hopes to improve the treatment of patients and increase awareness of this condition.

The Marfan Trust's three main objectives are:

1. To fund medical research projects that aim to aid the diagnosis and treatment of Marfan syndrome patients.
2. To provide educational literature on Marfan syndrome and all its aspects to the medical profession and the general public, thus raising awareness of the condition.
3. To provide peer-to-peer support and medical guidance to those affected and their families.

Our website includes information on the medical aspects of Marfan syndrome; advice and guidance on living with Marfan syndrome; updates on our latest research projects and publications and ways to support the Trust's important work.

The Sonalee Laboratory

In September 2000 the Sonalee Laboratory was opened at St. George's,University of London.

The laboratory was named after Dr Sonalee Wijetunge, a St. George's Hospital graduate, who died from Marfan syndrome.

The laboratory regularly receives samples from UK families and sometimes from all over the world, and it has achieved an enviable 91% success rate in mutation detection in classic Marfan syndrome.

The lab has access to an ABI Sequencing Genetic Analyser (3130xl), which enables the research team to screen patients and their families for mutations (gene errors) in the Marfan gene. Once a mutation has been identified, it can be used for diagnostic screening of at-risk family members, so that preventive therapy can be offered at the optimum time, thus improving the quality and quantity of life expectancy.

The effect of every mutation in the gene tends to be different for each family and for different patients in the same family. The lab is trying to make sense of this last statement.

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What is Marfan syndrome?

The syndrome is an inherited disorder of the body's connective tissue which predisposes to medical problems affecting the heart, eyes and skeleton, requiring treatment to prevent serious complications. It can affect men and women of any race or ethnic group. There are approximately 18,000 sufferers in the UK with around 200 new cases diagnosed each year. It is estimated that 1 in every 3,300 people are affected worldwide but that only half of sufferers are diagnosed.

Those affected suffer from one or more of the following problems:

Eyes: Dislocation of lenses, short-sightedness, retinal detachment, glaucoma

Skeleton: Excessive height with long limbs and fingers, flat feet, protruding or indented chest bone, loose joints, scoliosis, early osteoarthritis

Heart: Ballooning and potentially fatal tearing of the aorta, backward billowing of the heart's valves.

Sufferers can live with the first two problems – the last can cause death at an early age unless diagnosed in good time and treated medically and surgically.
Males and females are affected equally. Each child of an affected parent has a 50% chance of inheriting Marfan syndrome.

What causes Marfan syndrome?

A single abnormal (mutant) gene on Chromosome 15 causes the condition. This abnormal gene controls production of fibrillin, a very fine fibre in connective tissue throughout the body (the "glue and scaffolding of the body").

Most of the time this gene is inherited from a parent who is also affected. However, about 25% of the cases occur when the abnormal gene appears in an egg or sperm (a spontaneous "new" mutation) producing an affected child from two unaffected parents. Marfan syndrome is inherited as an "autosomal dominant" condition. This means that someone with Marfan syndrome has a 50-50 chance that each offspring will inherit the condition, regardless of sex.

How is Marfan syndrome diagnosed?

Marfan syndrome may be difficult to diagnose because signs of the condition vary greatly from one person to the next. Most affected people will not have all the signs and complications of Marfan syndrome. In general, Marfan syndrome is diagnosed after careful physical examination, particularly focusing on the main systems involved; eyes, skeleton, heart and lungs. Certain tests, such as an echocardiogram (a soundwave picture of the heart) are useful in making the diagnosis.

There is no cure for this condition, only diagnosis, monitoring and appropriate treatment.

To donate or get involved, go to the Marfan Trust website:

www.marfantrust.org

www.marfantrust.org/get-involved/donate

Or make a donation via bank transfer. Our bank details are as follows:

BANK: Charities Aid Foundation (CAF)

ACCOUNT NAME: The Marfan Trust

SORT CODE: 40-52-40

ACCOUNT NUMBER: 00017677

Or donate via the JustGiving page:

www.justgiving.com/marfantrust/donate

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