What is Marfan Syndrome?

Marfan syndrome (MFS) is a genetic disorder of the connective tissue, which helps to support many parts of the body. It was named after the French doctor who first described it in 1896. Marfan syndrome particularly affects the heart, blood vessels, skeleton and eyes. The disorder affects approximately 10,000 people in the U.K. and approximately 1 in 5,000 babies are born with MFS. It affects both men and women of any ethnic group. A number of famous people are thought to have had MFS including Abraham Lincoln, the American president and Paganini, the Italian violinist and composer. People with MFS are born with it; you cannot catch it.

What Causes Marfan Syndrome?

Marfan Syndrome is caused by changes in the FBN1 gene located on chromosome 15. This gene controls the production of fibrillin which is a very fine fibre that is found in connective tissue. Fibrillin fibres come together to form an elastic mesh which helps to support certain structures in our body such as blood vessel walls and the eye-lens. People with a changed FBN1 gene cannot produce enough fibrillin. The reduced amount of fibrillin causes medical problems which are described later on.

Most people with MFS (approximately 3 in 4) have inherited the changed gene from one of their parents who also has MFS. As some people with MFS may have a mild form of the disorder, this parent may not have been recognised until now as having MFS.

For a minority of people with MFS (approximately 1 in 4), the change in the gene has occurred by chance in the egg or the sperm that made them.

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HOW IS MARFAN SYNDROME INHERITED?

Marfan Syndrome is inherited in an autosomal dominant pattern. This means that every person with MFS has a 1 in 2 chance of passing the changed gene on to each of their children.

WHAT MEDICAL PROBLEMS ARE ASSOCIATED WITH MARFAN SYNDROME?

The severity of medical problems experienced by people with MFS can vary; some people are mildly affected whilst others have severe problems.

The most common problems are:

- **Cardiovascular (heart and blood vessels):** These are the most serious problems. In people with MFS, the aorta (the main blood vessel carrying blood away from the heart) tends to be wider and more fragile. If the aorta gets too wide, it can tear, leak and eventually rupture. This needs to be identified at an early stage so that it can be treated. It is also common that a particular valve in the heart (the Mitral valve) does not work properly. If left untreated, this can eventually cause heart failure.

- **Skeleton:** People with MFS are usually tall and thin with long arms, legs, fingers and toes. They commonly have loose joints which can be painful. They have a tendency to develop a curved spine, an abnormal chest bone, hernias and skin stretch marks.
- **Eyes:** People with MFS are often short-sighted. They may also suffer from a variety of problems with their eyes including their eye lens becoming dislocated.

**HOW IS MARFAN SYNDROME DIAGNOSED?**

Marfan Syndrome can be difficult to diagnose because the features of the disorder can vary so much from one person to the next.

**TO DIAGNOSE MFS, DOCTORS NEED TO ASSESS A PATIENT FULLY. THEY WILL TAKE A COMPLETE FAMILY HISTORY AND WILL CARRY OUT A FULL PHYSICAL EXAMINATION, AN ECHOCARDIOGRAM (AN ULTRASOUND SCAN OF THE HEART) AND A FULL EYE EXAMINATION. IN ORDER TO DIAGNOSE MFS, A MINIMUM NUMBER OF SIGNS IN DIFFERENT ORGANS OF THE BODY MUST BE PRESENT. DOCTORS USE A SET OF GUIDELINES CALLED THE ‘GHENT CRITERIA’ TO HELP THEM ASSESS A PATIENT AND DIAGNOSE MFS. HOWEVER, AS PEOPLE WITH MFS CAN HAVE A VARIETY OF FEATURES, THIS PROCESS MAY FAIL TO IDENTIFY THOSE WHO HAVE VERY MILD FORMS OF MFS.**

**IN PATIENTS SUSPECTED OF MARFAN SYNDROME ANALYSIS FOR CHANGES IN THE FBN1 GENE IS CURRENTLY AVAILABLE.**

**HOW CAN MARFAN SYNDROME BE TREATED?**

Unfortunately, there is currently no cure for MFS. Careful medical management can detect problems at an early stage. Together with an appropriate lifestyle, this will improve the outcome for people with MFS.

**Medical management** usually includes:

- **Regular echocardiograms to assess the heart and the width of the aorta.** Medication (Beta-blockers) can be given if the aorta starts to widen. This medication will lower blood pressure, slow the heartbeat and reduce stress on the aorta. If the aorta gets too wide, surgery may be needed to repair it.

- **Antibiotic prophylaxis may be required before minor surgery such as dental surgery.** This is recommended for people with MFS who have problems with the Mitral valve of their heart and by the advice of the cardologist. Antibiotics will help to reduce the risk of infection following surgery.
• Careful monitoring of the skeleton. This is particularly important during childhood and adolescence to detect problems with the spine or chest bone to treat them at an early stage.

An appropriate lifestyle for people with MFS usually means:

• Having a balanced healthy diet and avoiding smoking which is good for general health.

• Taking gentle exercise which is good for the heart, blood vessels and joints.

• Avoiding contact sports such as Karate to reduce the risk of damage to the aorta.

WHERE CAN I FIND MORE INFORMATION ABOUT MARFAN SYNDROME?

• The Marfan Association UK (Tel: 01252 810 472, www.marfan.org.uk) offers a wide variety of information leaflets and offers support to people with MFS and their families.

• The National Marfan Foundation in the U.S.A. (www.marfan.org) provides additional information and useful links to other websites.

Your local genetics service:

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Crewe Road South,
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