MARFAN SYNDROME

A Booklet for Teenagers

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INTRODUCTION

This booklet has been written especially for teenagers with Marfan syndrome.

- "What is Marfan syndrome?"
- "What affect will it have on me?"
- "Will I have to stop doing something I enjoy?"
- "What can I do instead?"
- "Why me?"

Keep reading the book – you are not alone!

It's sometimes difficult to get answers when you are a teenager. Well-meaning adults may think it is better for you just to have a little information, rather than a lot! Read on!

In actual fact it is important that **you** understand about Marfan syndrome, so that **you** can know how it is treated and how to interact with doctors and your family.

Having Marfan syndrome does not mean that you cannot have a full, exciting and rewarding life. The more you understand, the less it will interfere with your "hopes and dreams!"

YOU HAVE MUCH TO OFFER
AND YOU WILL ACHIEVE!

WHAT IS MARFAN SYNDROME?

Marfan syndrome is a disorder of the connective tissue that primarily affects the Eyes, Skeleton, Heart, Lungs, and Blood Vessels although it can affect other parts of the body too.

Everyone is affected differently and to varying degrees.

The eye problems may include short-sight (myopia) and possibly dislocated lenses.

In the skeletal system people with Marfan syndrome tend to be tall and thin, with long fingers, toes, arms and legs and the armspan often measures greater than height. The chest is usually narrow and may be abnormally shaped, either sticking out or in and the spine may have a curvature. Stretch marks (striae) may be present. You may have flat feet and unusually high arches, perhaps needing support. You may have a very high arch palate and need extra care at the dentist.

Your joints may be very flexible but

Do not be tempted to do "party tricks" by displaying what you can do, as it may cause you extra pain later in life!

Heart problems may include mitral valve prolapse, which is a floppy heart valve, and enlargement of the aorta, which is the main artery carrying blood from the heart.

Remember, help is available for all Marfan problems!

WHAT CAUSES MARFAN SYNDROME?

Marfan syndrome is a genetic condition caused by a change in one gene, present in every cell of the body. The gene causing Marfan syndrome can be inherited from a parent who also has the condition, or can occur only in the egg or sperm of an unaffected parent, commonly referred to as "a spontaneous mutation". *This just means the first in the family to be affected.* Approximately 75% of people with Marfan syndrome have a parent with the condition and 25% are the first to be affected.

The Marfan gene alters the connective tissue in the body, causing the typical changes in the eye, heart, skeleton and other organs. *Marfan syndrome can affect both men and women of any ethnic group.*

Each child of an affected parent has a 50% chance of inheriting Marfan syndrome and a 50% chance that they will not.

Marfan syndrome can vary widely in severity, even within one family, so a child with Marfan syndrome can be more or less severely affected than his or her affected parent.

THE MARFAN GENE WAS DISCOVERED IN 1990
ON CHROMOSOME 15q
Resulting from an international collaborative research study.

THE DEFICIENCY OF FIBRILLIN (part of the connective tissue structure) was discovered in 1991

TREATMENT

Early diagnosis combined with medication and appropriate physical activities can allow a more normal life span.

Remember you may have to see several doctors for the different body systems affected by Marfan syndrome. It certainly will be tiring and frustrating, but really is necessary!

An ophthalmologist (Eyes) may want to check your eyes annually, or more often, and may need to prescribe glasses, or contact lenses. Whilst some people with Marfan syndrome do not have any eye problems, many do.

You may find it helpful to read our Eye Pamphlet.

Cardiologist (Heart) Although it is unusual for young people with Marfan syndrome to feel ill due to heart problems, you should see a cardiologist regularly for check-ups. He will arrange an echo-cardiogram, which is a sound-wave test of the heart, to check on the size of the aorta and the functioning of the heart valves.

You may need beta-blockers, ace inhibitors or calcium antagonists to slow the rate and force of the heartbeat, minimising the risk of enlargement and tearing of the aorta. Enlargement is progressive and more likely to occur if the heart needs to work hard over a long period of time. The doctor will discuss the size of the aorta with you and your parents.

Competitive sports, vigorous physical work, or a blow to the chest (*as sometimes occurs in contact sports)* may cause a tear in the wall of the aorta (dissection).

Some people with Marfan syndrome, especially as they get older, will need heart surgery to replace both the portion of the aorta that is enlarged and possibly the valve between the aorta and the heart.

Elective surgery, that is surgery done at a chosen time, as opposed to an emergency, is now highly successful and has vastly improved the long-term outlook for people with Marfan syndrome. Ongoing checks will continue.

Skeletal System You will need ongoing evaluation by your GP, Paediatrician and Orthopaedic (Bones) specialist. Curvature of the spine can worsen, particularly during the adolescent growth spurt, when the rate of bone growth increases rather quickly. For some people, surgery may help to stabilise the spine, but this is not necessary for all Marfan patients. If you are especially concerned about the shape of your breastbone, you can discuss the possibility of surgery – but this is not recommended until growth is completed – and may certainly not be necessary.

IF YOU HAVE MARFAN SYNDROME
YOU ARE PROBABLY TALL AND ELEGANT
AND HAVE NO PROBLEM REACHING
HIGH SHELVES!

YOU AND YOUR DOCTOR

Mysterious Marfan

- We all hate going to the doctors, especially if we don't feel ill and if we expect to have tests or be given bad news.
- Keep informed about what is being done and why.
- Ask questions! Don't be frightened to take notes.
- Persist for answers!!
- You may feel more comfortable without your parents around for part of the consultation – if so, ask to spend some time alone with the doctor – then allow your parents to do the same!

HOSPITAL

- You may need to be admitted to hospital, either for surgery of for tests that cannot be performed as an outpatient. This can be frightening.
- Find out as much as you can ahead of time.
- Talk to your GP, your parents and your friends if this helps.
- Request a brochure about the hospital so that you know where you will be, what you need to take with you and visiting times.
- You may need to keep up with school/college work so check with your teachers about work being brought in to you
- You will meet lots of doctors and staff - ask questions! If they use medical language ask them to explain the words!
- Find out which doctor is supervising your case
- Before you leave, you may be given instructions

  concerning medication, diet, exercise
- Ensure you know what to do after leaving hospital and that you have relevant telephone numbers, appointment cards and medication

REACTION TO DIAGNOSIS

Some people are diagnosed as having Marfan syndrome when they are children. By the teenage years they will have learned how they are affected, what things are best avoided, which hobbies are most suitable – and, very importantly, what they enjoy most!

Sometimes parents believe they are offering protection by with-holding information and the teenager may feel resentful towards their parents and/or the doctors as he or she learns more about this connective tissue disorder. This is understandable as the teenager needs to understand and cope with the disorder. However, the teenager should remember that the parents may also find it difficult to talk about Marfan syndrome. Resentment may therefore be unjustified!

Some people are not diagnosed at an early age. Even before diagnosis many teenagers feel different from their friends. They are probably taller and slimmer, may need glasses and probably experience pain in various parts of the body, together with extreme fatigue and they may need to wear shoe supports. Many teenagers say they welcome the diagnosis as there is a reason for their "differences". Some say they feel "more whole" and "at one" with their friends, who usually become more supportive. Your reaction may be panic, fear, or anger at having to withdraw from a much loved sporting activity. You may hate being "labelled". Just remember that, whatever your reaction, it is fine, but do not "bury your head in the sand"

Learn to live life to the full - whilst protecting your health!

WHAT IS A WISE LIFESTYLE?

For the most part you can lead a near normal lifestyle. Some restrictions will be necessary on participation in contact sports and activities requiring excessive exertion, but not all sports are restricted. Competitive swimming may not be suitable but swimming itself is a very healthy sport. Such restrictions will be very hard for those who love and excel in sporting activities and may seem unfair. You are justified in feeling angry, but instead of letting it be destructive, develop confidence in other activities. Many teenagers excel in music and art. Long fingers and flexible joints are a distinct advantage when playing the piano, keyboard or guitar!

Talk to your doctor about which activities would be safe for you and discuss how sporting programmes could be modified.

Exercise regularly- eat a good balanced diet and remember that smoking is very bad for you!

One day you will find the person with whom you wish to share your life and that person will love you for yourself, completely accepting Marfan syndrome as being part of you. A loving and fulfilling sexual relationship can then be yours. At some stage you will both wish to discuss whether or not to start a family. As there is a 50/50 risk of passing on the Marfan gene to each child, you may wish to see a Genetic Counsellor at that time. If you are female it is wise to have a cardiac evaluation prior to pregnancy, due to strain being placed upon the heart and possible complications.

Many with Marfan syndrome do decide to have children!

TELLING OTHERS

It is up to you who you tell about Marfan syndrome but it is good to be open – having Marfan syndrome is certainly nothing to be ashamed of. In fact, it is impossible to find people who are not different in some way!

If you are not sure how to tell anyone,
give them this booklet and we will provide
you with another copy.
We can also send you a copy/copies
of our Marfan Fact Sheet.

Most people find it helpful to have a special friend who knows about Marfan syndrome and who knows what to do if a problem develops. This makes good sense!

CAREER PLANS

You will probably be thinking about your future – whether or not to continue with your education at college after leaving school or whether to find a job straight away.

Remember that the best education you can achieve
will bring you greater opportunity.

Do not feel that your choices are too limited as members of the Marfan Association UK work in the computing field, in music, as doctors, nurses, lawyers, teachers, writers, artists, secretaries and also in the media and in politics. The list is endless! In short, Marfan syndrome does not mean that you

will be denied a full and rewarding life. However, jobs that entail heavy lifting and great exertion are best avoided.

Explore your own special talents – you will have unique abilities – develop them to the full and, with determination, you can achieve much in your chosen career!

APPEARANCE

None of us are ever totally happy with the way we look!

Find out what styles and colours suit you best and develop your wardrobe. Being tall and slim can mean that certain styles look very good on you and you are probably the envy of your shorter friends!

Some physical features may worry you that are not at all obvious to your friends. However, appropriate styles can make you feel more comfortable.

If others sometimes are insensitive to you and make fun of the way you look, it is often because they are unhappy about themselves!

Although the adolescent years can be painful, as you get older you will find that your friends place more importance on your values, interests and ideas and looks do become less important.

You are far more attractive than you think!

FAMILIES REACTION TO DIAGNOSIS

Each family reacts in a different way but the whole family is affected by the diagnosis and not just the affected person.
In many families a parent is also affected. Anger or blame may be directed at the affected parent, which can then lead to a feeling of guilt on both sides. This is normal! Bring it into the open and you will find that you can share experiences.
You may feel jealous of unaffected brothers or sisters as they do not need to restrict activities, or go to the hospital regularly, or take medications. They may actually be jealous of you, thinking that you get more attention than they do! It will help you all to discuss openly.
You may think your parents are "taking over your life". They are only concerned. Take your medication before they remind you and they will realise that you are becoming responsible for yourself. They will feel less anxious and you will feel that you are in control!

LIVING WITH MARFAN SYNDROME

You will experience many emotions – anger, fear, depression and exasperation. If a parent is affected, you will be worried for them too. Life's restrictions will be a nuisance. Trying to buy clothes and shoes to fit can be frustrating and tiring. You may feel alone – but you are not! Your family and friends love you – share your questions and feelings with them and life will become easier. Learn about Marfan syndrome, have regular check-ups but then get on with your life!
You're a person 1st and a person with Marfan syndrome 2nd!