Guide to Physical Activity

Reviewed by Dr Sabiha Gati, Consultant Cardiologist, Royal Brompton Hospital

Cover Star: Marfan Trust supporter, Freddie Beard

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Marfan Trust, a CIO registered as a charity in England in Wales with charity number 1198847

What is Marfan Syndrome?

Marfan syndrome (MFS) is a genetic disorder of the body's connective tissue that affects any gender, race or ethnic group. Connective tissue helps provide structure to the body, binding skin to muscle and muscle to bone. It provides the stretchy strength in tendons and ligaments around joints and in blood vessel walls. It also supports the internal organs. This tissue is made of fine fibres and 'glue'. One protein in these fibres is called fibrillin.

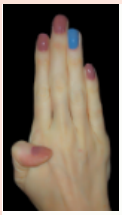
In MFS, a change in the fibrillin-producing gene, fibrillin-1, means that this protein is deficient in connective tissue throughout the body, creating an unusual stretchiness and weakness of tissues. This has far-reaching implications and can affect the eyes, lungs, gut, nervous system, skeleton and, most dangerously, the cardiovascular system. Symptoms can vary widely from person to person with people experiencing mild to severe manifestations.

MFS affects roughly 1 in 3,000 people which means that approximately 18,000 people in the UK have MFS. We estimate that half remain undiagnosed. 75% of patients inherit the condition whilst 25% develop it as a result of a spontaneous (new) gene change. Each child of an affected parent has a 50% chance of inheriting Marfan syndrome.

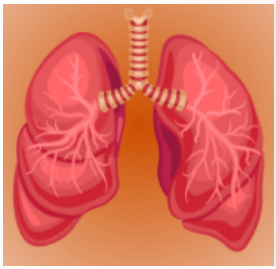
Every year, on average in the UK there are over 200 new cases of Marfan diagnosed. Loeys-Dietz syndromes are rarer, have some overlap in signs and symptoms, and much of the exercise advice applies equally to them.

Salient Features of the Syndrome

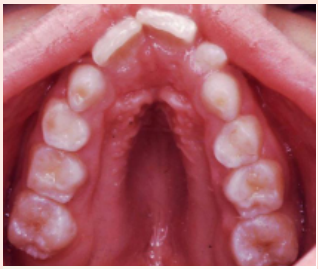
Marfan syndrome was first identified in 1896 by French physician, Dr Antoine Marfan. It is a multi-faceted disorder which can be difficult to diagnose because signs of the condition vary greatly from person to person. However, there are salient features which we outline below:



Skeletal: Musculoskeletal problems are common and troublesome in MFS, with patients often growing to excessive height whilst developing curvature (scoliosis/kyphosis/lordosis) of the spine. Fingers are often spidery and long, and hammer toes are a frequent feature. Skeletal problems also involve abnormally shaped chest (pectus deformity), and loose joints which often cause pain and occasionally, joint dislocation.

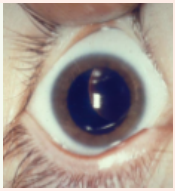


Respiratory/Lungs: Pulmonary complications occur in 10% of patients, commonly cystic changes in the lung, and occasionally pneumothorax (air trapped outside the lung due to an air leak). Further complications can include bronchiectasis, fibrosis, emphysema and asthma.



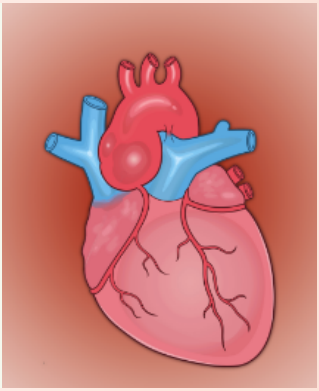
Dental: A high-arched palate and crowding of teeth with a history of tooth extraction for crowding is fairly common amongst Marfan patients.

Eyes: Ocular manifestations of MFS include myopia (short-sightedness), with some experiencing subluxation or dislocation of the ocular lens (ectopia lentis). Strabismus (squint) and glaucoma are also symptoms, as is retinal detachment.



Heart: The most serious problems occur in the heart and blood vessels. People can experience ballooning and potentially fatal tearing of the aorta (the body's largest artery, carrying oxygen-rich blood from the heart to the circulatory system) and backward billowing of the heart's valves. Left unmonitored or untreated, these symptoms can be life-threatening.

The key to a long and healthy life with Marfan syndrome is monitoring with regular scans to check the heart function and the size of the aorta. A combination of echocardiograms and MRI or CT scans is usually used. This allows doctors to plan any treatment that may be required.



Introduction

A reliable producer of happy hormones, regular physical exercise elevates the mood, helps health and drives energy. It has proven benefits for our physical and emotional wellbeing and can be safely integrated into the life and daily routine of someone with Marfan syndrome.

Until recently, there was an emphasis within healthcare settings on what people with Marfan syndrome cannot do, rather than what they can. This negative approach is rapidly changing as the physical and psychological advantages of recreational exercise become increasingly apparent.

Regular (gentle) physical activity can help to counter and alleviate the features of MFS that worsen with age - chronic fatigue and joint problems, for example. Adapting protective physical measures into your lifestyle will help to reduce health concerns.Yet there is an exercise paradox for people with Marfan and Loeys-Dietz syndromes in that it can be hazardous.

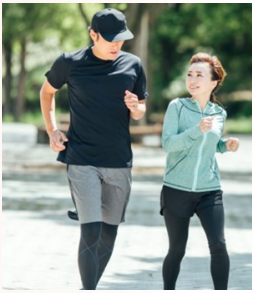
Tailored, specific advice from a healthcare professional is essential

Every person with Marfan syndrome is affected differently: whilst it's possible to offer some general guidelines, it is vital that everyone is treated as an individual with their specific risk factors taken into consideration. Connective tissue is our 'scaffolding' and can be found throughout the body. The signs and symptoms of MFS can therefore appear anywhere, from the heart, eyes and lungs to the musculoskeletal system. Features of MFS can even appear in the skin. Some of the physical manifestations of the syndrome may limit the person's ability to fully participate in exercise. For example, those with eye problems may have difficulty with sports involving hand-eye coordination, such as racquet sports. People with Marfan syndrome are affected in different ways, so what is suitable for one person may not be for another. If the aorta is affected, they should ideally avoid contact sports.

Instrumental in helping the individual decide what is best for them is the healthcare professional who will offer tailored, specific advice about the types of activity patients can safely perform. It is essential for a person with Marfan syndrome to discuss physical activity and the specific levels of activity with their physician before embarking upon an exercise regime. Shared decision-making between the clinician and the patient is key.

Different Types of Exercise

- **Aerobic**: aerobic means 'with oxygen' and describes 'cardio' exercise. It is when you're moving your body in a way that makes you warm and slightly out of breath. Swimming laps, walking, cycling, housework or gardening constitute aerobic exercise. Whilst engaging in this dynamic, continuous activity, your heart rate increases, and your body's cells use oxygen to produce energy. Over time, this helps your heart,lungs and circulatory system to work better by lowering the blood pressure and resting heart rate, improving cholesterol levels and helping you to maintain a healthy weight.



By contrast, Anaerobic means 'no oxygen'. This is a higher intensity, higher power version of exercise such as sprinting, weightlifting, biking, skipping rope and high-intensity interval training. This uses glucose, not oxygen, for 'fuel' and is performed in bursts. It can be hard on the body.

- **Balance, Stretching and Flexibility**: exercises including yoga, tai chi and Pilates where we hold our bodies in less stable positions ensure that our muscles don't get too tight and keep us flexible, helping us to avoid pain or injury while reducing the risk of having falls.

- **Resistance and Strengthening**: these exercises may become isometric and static, and include lifting weights, body-building or using **resistance bands** and cables to strengthen your muscles. The stronger your muscles, the harder they can work, which takes the strain off your heart, making it easier to do everyday tasks. But this comes with caveats.

When these exercises involve strenuous straining, pushing and pulling, they can lead to a sharp rise in blood pressure, thereby putting a strain on your aorta which is to be avoided for anyone with Marfan syndrome.

Many physical activities involve a combination of the above, mixing energetic and static muscle work with aerobic and anaerobic energy use. Generally speaking, exercise and physical activity that involves movement at a comfortable pace (during which a normal conversation be held) is safe. Exercise that leads to straining, pushing, and bearing down at high resistance or to levels of exhaustion may potentially cause injury and should be discussed with your clinician.

Marfan Syndrome Concerns – the Three Cs: Collision, Competition & Contact

Whilst regular, recreational exercise improves everyone's wellbeing, when taken to an elite or competitive level it can harm the health of people with Marfan syndrome. Contact sport and anything involving potential collision is also to be avoided. These are the risks to be taken into consideration during physical activity:

- **Eyes** in Marfan syndrome are vulnerable in a contact sport. An impact to the head can be damaging, worsening/risking lens dislocation or leading to retinal detachment. Short-sightedness can affect co-ordination and may increase the risk of collisions. Contact sport should be avoided.
- **Bones & Joints** any excessive stress and strain can induce joint pain and injury. Height and long limbs may also affect hand-eye co-ordination while lack of muscle bulk can lead to fatigue at an earlier stage than for the average person;
- **Heart** spikes in blood pressure and straining can add stress to an already-dilated aorta. The aorta should remain protected from external force or injury;

Medications: beta blockers work to lower blood pressure and reduce the workload of the heart. In doing so they reduce the heart rate. Whilst beta blockers do not reduce the benefits of exercise they can mean your heart rate remains slower and does not respond to exercise in the same way as it did previously. You may therefore need to modify your exercise regime until you are used to your new medication. Some people with with Marfan and Loeys-Dietz syndromes who have had heart valve surgery may be taking lifelong anticoagulants which increase the risk of bleeding. Contact sports should be avoided when on these medicines.

The table below delineates a spectrum of physical activities and their safeness, or otherwise, for people with MFS. Each patient is affected differently, and the table therefore denotes which of the systems is placed at risk by which sport.

Contact, Collision & Strenuous	Boxing (E, H) Basketball (E, H) Handball (E, H) Martial Arts (E, H)	Rugby (E, H) Water skiing (E, H, J) Wrestling (H, J)
Contact Limited but Strenuous	Football (E, H) Gymnastics (E, H) Hockey (E, H)	Horseback Riding (E, H) Squash (E, H) Volleyball (E, H)
No Contact but Strenuous	Cycling (intense H) Skiing (downhill) (H) Weight-Lifting (H)	Running (H)
Atmospheric Change Activity	(A change in air pressure can lead to pneumothorax, collapsed lung)	Deep-sea Diving Scuba Diving Sky-diving
Recommended Sport (not at high-level competition)	Archery Badminton Bowls Canoeing Cricket Cycling (on the level) Discus Dancing Golf Javelin	Netball Raquetball Sailing Skating Swimming Shot-put Table Tennis Tennis (doubles) Yoga Walking/Jog-Walking

(Modified from Marfan Foundation US Physical Activity Resource)

Key: E – Eyes; H = Heart; J = Joints

Exercise & Aortopathies

In 2020, the European Society of Cardiology published a new set of guidelines on sports cardiology and exercise in patients with cardiovascular disease. While acknowledging that Marfan, Loeys-Dietz and Ehlers-Danlos syndromes can confer a much higher risk of an aortic event at a much younger age during intensive exercise, the guidelines recommend physical activity for all patients with aortic pathology. Within the guidelines is a focus on shared decision-making between clinician and the patient who wants to exercise.

Amongst many studies underpinning the latest thinking on the benefits that exercise brings to an MFS patient is one that examined the effects of physical activity on the aortic diameter and wall stress of a Marfan animal model. The mouse was made to run on a treadmill with a mild to moderate dynamic intensity. This resulted in a reduction in the growth rate of their aortic diameter whilst their aortic wall became stronger. The optimum protective effect was a training intensity of 55 – 65% of peak VO_2. This translates to mild-to-moderate intensity exercise. (VO_2. is a measurable value of oxygen consumption from a session of physical exercise). Here is a useful table categorising various physical activities and their corresponding intensity level.

Meanwhile, a recent randomized controlled study explored the benefits of a 3-month online supervised training program for patients with Marfan Syndrome (MFS). This French study from VASCERN found:

Key Findings:

- Quality of life improved by +20%.
- Peak oxygen uptake increased by 34%.
- Significant improvements in muscle elasticity and blood pressure.
- Importantly, no change in aortic root diameter, ensuring safety for MFS patients.

This study suggests that online personal training could be part of a management strategy for MFS -

Here is a useful table categorising various physical activities ...

Low-Intensity, Light Exercise	Moderate Intensity Exercise	Vigorous Exercise
Walking (leisurely) **Sitting** (desk work) **Light housework** (dishes, sweeping) **Fishing** **Playing a musical instrument** **Gardening** (light) **Golf** (with cart) **Boating** **Sailing** **Bowling**	**Walking** (briskly—4 mph) **Heavy Cleaning** (mopping, vacuuming) **Mowing lawn** (using power mower) **Bicycling** (leisurely pace—10-12 mph) **Dancing** (leisurely pace—ballroom) **Badminton** **Golf** (pull cart, walking) **Doubles tennis** (leisurely) **Yoga** **Pilates** **Water aerobics** (leisurely) **Swimming** (recreational, light) **Calisthenics** (light, without weights) **Raking lawn**	**Handball** **Hiking** (moderately up steep grade) **Jogging moderately** (6 mph) **Farming** (hay bailing) **Singles tennis** **Basketball** **Soccer** **Bicycling at a fast pace** (14-16 mph) **Swimming** (fast) **Football**

(Modified from Marfan Foundation US Physical Activity Resource) and Source: **https://www.cdc.gov/mmwr/preview/mmwrhtml/mm5009a3.htm**

How to Exercise Safely

Woven throughout this Guide is an important refrain: it is essential for a person with Marfan syndrome to discuss physical activity and the specific levels of activity with their physician. The importance of this cannot be over-emphasised. For those with aortic involvement, it is advisable to organise a risk assessment prior to embarking on a new regime, i.e. a careful examination with advanced imaging of the aorta (CT/CMR) and exercise testing with a blood pressure assessment.

- When choosing your sport, it's safe and indeed highly advisable to pursue a non-competitive recreational sport at a low towards moderate intensity and low impact – one that can be easily integrated into everyday life, say 30 minutes five or six days a week. If this is too much, then, shorter bursts.
- Taking beta blockers will slow the heart rate and reduce the blood pressure. The pulse rate therefore becomes less responsive to exercise, so it is harder to make rapid changes to the activity level, and the pulse rate is a less reliable way to assess your level of exertion. This means that you may not meet your ideal level of fitness.
- If you can talk in a conversational tone of voice during the activity, you are probably exercising at a reasonable level. If you are taking a beta-blocker, try to keep your pulse to 60% of maximum age predicted (60% of 220 minus your age e.g. for a 50 year old, 220 – 50 equals 120 and 60% is 102 beats per minute), or try to keep the heart rate from increasing more than 30% from baseline. (It is often easier to feel the pulse over arteries in the neck than at the wrist.)
- Do not test your limits.

Advice for Children

Intrinsic to a child's happiness is a sense of belonging. It's an instinctive human need, providing security and stability. Sport nurtures and builds a feeling of belonging, fostering inclusion, while teaching children how to play with and compete against each other.

The symptoms of Marfan syndrome may prohibit a child from participating in certain sports at school, rugby or handball, for example. This may leave them feeling isolated and different. *It's therefore important to integrate the child in a different but important way, perhaps by appointing them referee or umpire.*

Some may be aspiring sport journalists and so could become the official commentator. Assigning a tangible task to a child who could feel otherwise excluded is key to retaining a feeling of inclusion.

The considerations during physical activity are the same as those for adults, namely the heart, bones and joints, and eyes. As with adults, children with MFS should be encouraged to choose low-impact, low-intensity activities and their expectations need to be managed: participating in sports at an elite or competitive level will not be possible and this can be difficult to hear, particularly if their diagnosis is made at a later age and they are already playing or competing at a high level.

A useful first step can be for schools to provide parents with information about the PE/ sporting curriculum for the coming academic year so they can obtain specific, tailored advice from the doctors.

The age of diagnosis will impact the child's response to restrictions on their physical activity. If they have a pre-school diagnosis, their activities can be tailored from an early age and other activities can be incorporated, e.g. hobbies such as art, music and photography can foster a community and a social circle that is common within sport. If a child is diagnosed at an older age, they may already be participating in sports at a high level and be a valued member of a team that is an important part of their identity and social circle. This is challenging and can lead to them feeling excluded, angry and depressed if they can no longer be a part of this team. This requires careful management and can be incredibly difficult for a teenager.

Echoing our advice for adults, children should:

- **Avoid contact sports, high-level competitive sports and static/ isometric exercise in which the muscles and blood vessels contract, causing spikes in blood pressure.**
- **Try to maintain an aerobic level of activity (the child should be able to continue to hold a conversation whilst exercising).**
- **Light weights can be used with higher numbers of repetitions rather than heavy weights with fewer repetitions**
- **Wear protective equipment e.g. bike helmet, squash goggles.**
- **Incorporate adequate time for warm-up and cool-down.**
- **Avoid 'fitness tests' that pit children with MFS against other children; try to focus on personal achievements and achieving a 'personal best'.**
- **Activities such as football, basketball, cycling and dance are all highly strenuous but can also be enjoyed.**
- **Regular physical activity is important and children with MFS should be encouraged to find a sport or activity that they enjoy.**

Final advice should always come from the child's own doctor.

What to Do While Waiting for a Diagnosis

Awaiting a diagnosis is an anxious time. Everything feels upended as you are suspended in temporary limbo. Should you continue life as normal, or adapt your activities in anticipation of a diagnosis that may prove to be negative?

Assuming you are already competing in a contact sport, or pursuing a strenuous exercise regime, it is advisable to temporarily stop. It is also advisable (if this hasn't already happened) to seek an echocardiogram whilst you await the test results.

Ideally you should seek advice from your doctor – GP, cardiologist, optometrist.

Tips from the Top: Innate Advantage

Constraints inspire creativity, and limitations can be turned into advantages. Marfan patients are often reminded of their physical vulnerabilities and warned against certain sports. But what of those activities that can exploit Marfan attributes? After all, height and limb length confer excellent reach.

Dr Sabiha Gati, cardiologist at the Royal Brompton Hospital says: *"Archery is really good for Marfan patients as it helps to build the upper arm muscle strength, and it's conducive to the Marfan frame. You need slightly stronger upper arm muscles for this sport, and so archery helps to not only build on that aspect but also on one's stature and physique. It also enhances your posture. This is so important. You can use your natural assets – your long arms - to your advantage in archery."*

Cover Star

We leave the last word to Freddie, a Marfan Trust supporter and this guide's cover star:

"My name is Freddie Beard, I have been diagnosed with Marfan syndrome since 2014 thanks to a chance meeting with a junior doctor who was sitting in on a regular doctor's appointment. The junior doctor had recently been studying Marfan Syndrome, and noticed that my pigeon chest could in fact be part of this. Fast forward to 2016, and after having all the tests to confirm it, I was in need of open heart surgery to correct my leaking aortic valve. The surgery was very intense, scary, and worrying, but the doctors at Bart's Hospital were fantastic and were able to lead to me being here today.

Their fantastic work has allowed me to be able to rediscover my love of swimming after not being too sure how I would be able to keep active post-surgery. Instead I was able to pick up both swimming and water polo six months after surgery and I've been able to continue swimming ever since! Swimming has helped me rediscover the freedom and confidence in my own body when exercising, be that in a pool, or when swimming in open water. It gave me a physical goal to aim for, and continues to fit well with Marfan and the frame it has given me. If anything, having a longer wing span of arms due to Marfan Syndrome surely makes me even quicker than your average swimmer!

Marfan fits so well with my swimming life style that I have been able to complete The Hyde Park Serpentine swim for two miles, and most recently for the six-mile distance! So all in all, Marfan alongside my swimming has really kept me afloat (pun intended!) both physically and mentally."

Glossary of Terms

Aortopathy is a condition that weakens your aorta

Bronchiectasis is a chronic lung condition where the walls of your airways (bronchi) widen and are thickened from inflammation and infection.

Emphysema is a lung condition that causes shortness of breath. In people with emphysema, the air sacs in the lungs (alveoli) are damaged. Over time, the inner walls of the air sacs weaken and rupture — creating larger air spaces instead of many small ones. This reduces the surface area of the lungs and, in turn, the amount of oxygen that reaches your bloodstream.

Isometric Exercise denotes a muscular action in which tension is developed without lengthening the muscle.

Isotonic Exercise occurs when the tension or torque generated by the muscle is constant throughout the movement. In practice this is very difficult to accomplish, and a better term is dynamic contraction, which may be subdivided into several types of contractions: concentric, eccentric, and isokinetic.

Kyphosis is a curvature that makes the spine more rounded and can make a person appear hunched over.

Lordosis is an exaggerated inward curve of the spine that typically affects the lower back, a condition called lumbar lordosis. Less often, children develop a pronounced inward curve of the neck, cervical lordosis. Lordosis can affect people of all ages. **Pneumothorax** is a collapsed lung.

Scoliosis - describes spinal curvature side to side, with twisting.

VO_2 peak is a measurable value of oxygen consumption from a session of physical exercise.

Further Resources:

2022 ACC/AHA Guideline for the Diagnosis and Management of Aortic Disease: A Report of the American Heart Association/ American College of Cardiology Joint Committee on Clinical Practice Guidelines

https://www.ahajournals.org/doi/ 10.1161/CIR.0000000000001106

About the Marfan Trust

Co-founded in 1988, the Marfan Trust is the sole charity in the United Kingdom dedicated to improving and saving the lives of those with Marfan syndrome. It is estimated that approximately 18,000 people are living with Marfan syndrome in the United Kingdom and around half of these remain dangerously undiagnosed.

The Marfan Trust's three main objectives are to:

- Provide personalised support and medical guidance through its helpline;
- Conduct cutting-edge medical research through its self-funded Sonalee Laboratory, named after a young doctor who tragically died of complications from MFS during her ward round;
- Continue to provide educational information and raise awareness of the condition.

How you can help

Marfan Trust – Supporting Research into Inherited Aortic and Related Disorders

Loeys Dietz

You can help to secure the Marfan Trust's future by becoming a member today for just £3 per month:

www.marfantrust.org/pages/10-membership

JustGiving

Just Giving – **http://bit.ly/3Scj51w**

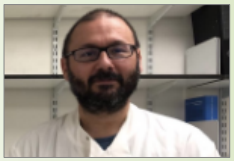
PayPal

PayPal Giving – https://bit.ly/45NCuwQ

By donating to the Marfan Trust, you are contributing to an ever-growing body of knowledge on the condition, allowing more doctors and medical specialists to deliver the best possible treatment to patients affected by Marfan syndrome.

- BANK: Charities Aid Foundation (CAF) ACCOUNT NAME: Marfan Trust
- SORT CODE: 40-52-40
- ACCOUNT NUMBER: 00017677
- REFERENCE: Your Name (plus campaign name if relevant)

You can also help to fund a piece of equipment in Dr José Aragon-Martin's Sonalee Laboratory. Email info@marfantrust.org to find out more.



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