

Marfan Trust
Supporting Research into Marfan Syndrome



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Exercise Guide

Marfan Syndrome

THIS LEAFLET IS WRITTEN BY:

DR ANNE CHILD MD FRCP AND BETHAN DAVIES MBBS

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www.marfantrust.org

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What is Marfan syndrome?

An inherited disorder of connective tissue that affects many organ systems including the skeleton, lungs, eyes, heart and blood vessels. This condition can affect both men and women of any race or ethnic group. It is estimated that around 18,000 people in the United Kingdom have Marfan syndrome.

Salient Features

Skeletal: typically tall thin physique, with long limbs and fingers, scoliosis, narrow chest with pigeon or funnel deformity, joint hypermobility and dislocations. Dural ectasia occurs in 65% of patients.

Cardiovascular: dilatation of ascending (and sometimes descending) aorta, incompetence of aortic and mitral valves, aneurysm and dissection of aorta.

Respiratory: pneumothorax, bronchiectasis, emphysema and asthma.

Ocular: subluxation or dislocation of lens, myopia and unstable refraction, detachment of retina, strabismus, glaucoma.

Dental: high arched palate, crowding of teeth.

Genetic: males and females are affected equally frequently. Each child of an affected parent has a 50% chance of inheriting Marfan syndrome. In 25% of cases neither parent is affected; however, apparently unaffected parents should be screened carefully as the severity and pattern of disease are variable, even within one family.

Diagnosis

Diagnosis is made after careful physical examination and echocardiography, demonstrating classical features in 2 out of 3 major systems (eyes, heart, skeleton), supported by a family history in 75% of cases. Mutations can be found in the fibrillin-1 gene in 97% of patients, assisting with screening of family members, and pregnancies. Diagnosis can also be confirmed within a family by genetic linkage studies.

Cardiac problems

The most serious problems occur in the heart and blood vessels. The aorta is usually wider than expected for a given body surface area and is more fragile due to a deficiency in the amount of Fibrillin present. The dilatation tends to be progressive leading to aortic dissection +/- aortic regurgitation. Surgical repair is recommended when the aortic root becomes widened to 4.8cm, or earlier in cases with a family history of early dissection. Beta-blocker therapy can delay dilatation. Mitral valve prolapse is also often present. Antibiotic prophylaxis is recommended for dental procedures involving bleeding, if a heart valve is leaking sufficiently to produce an audible heart murmur.

Introduction

For people with Marfan syndrome, certain activities are thought to increase the risk of some of the serious complications. In addition, some of the physical manifestations of Marfan 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.

Guidelines

The Marfan Trust www.marfantrust.org provides guidelines on what exercise is suitable for people with Marfan syndrome, and which activities are best avoided. There is currently no evidence base for how exercise affects people with Marfan syndrome and therefore the advice given should be based on the guidelines issued by the Marfan Trust.



Recommended (Not high level competition):		
Archery	Cycling (on the level)	Hockey
Shot-put	Badminton	Discus
Javelin	Skating	Bowls
Fencing	Netball	Swimming
Canoeing	Football (no heading)	Racquetball
Table Tennis	Cricket	Golf
Sailing	Tennis	Yoga
Walking/jog-walking	Dancing	Light weight-lifting

Please remember each patient is affected differently and our general recommendations need to be discussed with the patient's own medical attendants and parents. Children should be allowed to stop when tired.



Contraindicated: to Prevent Damage to:			
Boxing	E	*Squash	E H
Rugby	H J	High Altitude Mountaineering	L
Deep Sea Diving	L	Trampolining	J
Rowing	H J	High Diving	E
Distance Running	H J	*Weight-Lifting	H J
Sky-Diving	L	Karate/Judo	H J
Hang Gliding	L	Wrestling	H J

* Key: **E** = Eye | **H** = Heart | **J** = Joints | **L** = Lungs

H: Heavy lifting is not advised.

H: Basketball should be discussed with the patient's doctor. If played as a contact sport, it can lead to repeated falls.

H,J: Horse riding on a quiet horse is best. Jumping may cause falls.

E, H: *Squash : for those who do not have heart problems, squash can be played wearing goggles to protect the eyes.

H, J: Participating in Aerobics and Abseiling should be governed by the patient's limitations.

Prolonged exertion at peak capacity should be avoided. In the gym, short stints in a number of activities are recommended.

For children, alternative activities could include swimming in the school pool, to be undertaken at the same time as the peer group is performing more stressful PE activities. If that is impossible then the child should be given a task, with the necessary instruction to enable the child to complete it competently, such as refereeing which gives them a position of importance without being involved in the physical activity. Some children are given an individual fitness routine to work through in the corner of the gym.

Final advice should come from the patient's own doctor.

The Problems Encountered by Patients

In a survey on the **exercise habits** of 70 patients aged between 18 and 65 with Marfan syndrome, the following results emerged:

- ♦ 72% said that their exercise habits had been affected by having Marfan syndrome.
- ♦ The reasons for this were (most common first) joint pain, shortness of breath, medical advice, fatigue, palpitations, dislike of exercise, and overheating.
- ♦ By far the most popular exercise activity was walking, followed by gym, swimming, jogging, cycling and yoga/pilates. Only a handful of people participated in activities that are considered to be unsuitable (long distance running, heavy weight-lifting).
- ♦ Only 44% of people had received information about exercise.
- ♦ Of these, the most common source of information was a hospital consultant, followed by the Marfan Trust, GP, the internet and occupational therapists/physiotherapists.
- ♦ The advice given seems to have corresponded with the published guidelines.
- ♦ 17% said that they had received conflicting advice.
- ♦ Of the 25 people who had cardiac surgery, only 13 had been offered subsequent rehabilitation. Of these, 9 took up the offer and in only 4 cases was the person in charge of the rehab definitely aware of Marfan syndrome.
- ♦ A number of those engaging in rehab found that it was not tailored to their needs, but to the needs of an older population with different conditions (e.g. coronary artery disease).

Other Findings

Many people commented that, although they had been given general exercise advice, they were still unclear about what it actually meant in practice. In addition, they often found that doctors, other healthcare professionals and employees of gyms/fitness centres were not able to give accurate and appropriate advice.

Worryingly, 2 people had been turned away from gyms once they disclosed that they had Marfan syndrome. A letter from the doctor to the gym instructor indicating suitable sports is therefore recommended. Some found the exercise advice frustrating and demoralising; for those who were keen to improve their fitness, positive suggestions on how to achieve this were limited – much of the advice focussed on what not to do.

A number of people commented on their negative experiences of sport at school and the distress at being told that they could no longer do their favourite sport or compete. Tapering unsuitable sports such as long distance running, and gradually replacing them with more suitable sports is recommended.

Who should be aware

Hospital Doctors, GPs, physiotherapists and those working in the fitness industry should be aware of the nature of Marfan syndrome and how it relates to exercise ability. A letter from the Hospital Doctor to a physical education teacher or other supervisor of exercise programme is helpful.

What advice is needed

A system should be in place to ensure that all people with Marfan syndrome receive information about exercise as soon as possible after diagnosis. Patients should be encouraged to exercise as much as possible within their capabilities in order to improve and maintain their general health.

When giving advice to patients, the advice should be tailored to the needs of the individual, taking into account how they are affected by Marfan syndrome, their age and their general health.

Patients need more than a couple of activity suggestions (such as walking and swimming). They need to know what level they should exercise at, how long and how often to exercise and, especially for those keen to maximise their fitness, practical suggestions on how this can be achieved. Where possible, the patient's own preferences of sporting and exercise activities should be accommodated.

For those who are involved in exercise considered to be a risk to their health, the advice should be to taper down the activity over a period of time, rather than stopping it suddenly. This is more acceptable to the patient and gives time to develop an interest in an alternative sport. As a general rule, the patient should be able to converse while exercising.

After Surgery

Following heart surgery, patients should be offered a cardiac rehabilitation programme suitable to their needs and age.



Marfan Trust | Registered Charity No. 328070

Dr. Anne Child MD FRCP Medical Director, Marfan Trust

Guy Scadding Building

Dovehouse Street

London | SW3 6LY

T: 020 7594 1605

www.marfantrust.org | E: info@marfantrust.org

