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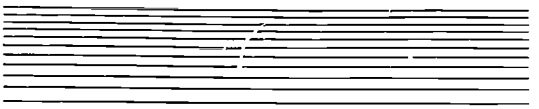
ABSTRACT

This document consists of two brochures, the first explaining the Marfan Syndrome and a second providing guidelines for physical education and activity for people who have this syndrome are provided. The brochure on factual information about Marfan syndrome outlines the associated medical problems involving the cardiovascular system, the skeleton, and the eyes. Also covered are causes and how to diagnosis and treat this disorder. The functions of the National Marfan Foundation are outlined. The brochure on physical activity presents information on changes in connective tissue that cause the Marfan syndrome, differences among forms of exercise and competition, and considerations when medications are being taken. Guidelines and modifications are included to promote safer exercise for people with the Marfan syndrome. A chart classifies sports and activities into four categories: strenuous and contact/collision high potential, strenuous and limited contact, strenuous and noncontact, moderately strenuous and noncontact, and nonstrenuous and noncontact. (SW)

The Marfan Syndrome. Fact Sheet [and] Physical Education and Activity Guidelines

THE MARFAN SYNDROME



NATIONAL
MARFAN
FOUNDATION

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FACT SHEET

What is the Marfan syndrome?

The Marfan syndrome is a heritable disorder of the connective tissue that affects many organ systems, including the skeleton, lungs, eyes, heart and blood vessels. The condition affects both men and women of any race or ethnic group. It is estimated that at least 40,000 people in the United States have the Marfan syndrome.

What medical problems are associated with the Marfan syndrome?

The Cardiovascular System

- The most serious problems associated with the Marfan syndrome involve the cardiovascular system. The two leaflets of the mitral valve may billow backwards when the heart contracts (mitral valve prolapse). This can lead to leakage of the mitral valve or irregular heart rhythm.
- In addition, the aorta, the main artery carrying blood away from the heart, is generally wider and more fragile in patients with the Marfan syndrome. This widening is progressive and can cause leakage of the aortic valve or tears (dissection) in the aorta wall. When the aorta becomes greatly widened, or tears, surgical repair is necessary.

The Skeleton

- Skeletal manifestations common in people with the Marfan syndrome include curvature of the spine (scoliosis), abnormally shaped chest (pectus deformity), loose jointedness and disproportionate growth usually, but not always, resulting in tall stature.

The Eyes

- People with the Marfan syndrome are often near-sighted (myopic). In addition, about 50 percent have dislocation of the ocular lens.

How is the Marfan syndrome diagnosed?

The Marfan syndrome is difficult to diagnose because there is no specific laboratory test for the condition. In addition, characteristics of the disorder vary greatly among affected individuals. Most affected people do not have all of the possible signs and complications of the syndrome.

An accurate diagnosis of the Marfan syndrome can be assessed after a complete physical examination that focuses on the systems affected by the disorder. This includes:

- Echocardiogram, a sound wave picture of the heart by a cardiologist.
- Slit-lamp eye examination by an ophthalmologist.
- Skeletal examination.
- Complete family history.

The recent identification of the chromosome, gene and component of connective tissue (fibrillin) in which the mutation for the Marfan syndrome is located offers great promise for the diagnosis of the condition. It is hoped that as a better understanding of fibrillin is gained, earlier and more accurate diagnosis of the Marfan syndrome will be possible.

How is the Marfan syndrome treated?

People affected by the Marfan syndrome should be treated by a physician familiar with the condition and how it affects all body systems. There is no cure for the disorder yet, but careful medical management can greatly improve the prognosis and lengthen the life span.

Every affected person should work closely with his/her physician(s) for their customized treatment plan. However, in general, treatment includes the following:

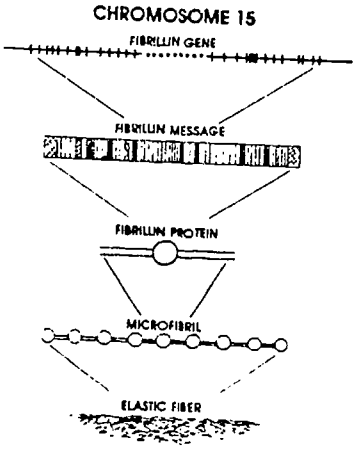
- Annual echocardiogram to monitor the size and function of the heart and aorta.

- Initial eye examination with a slit-lamp to detect lens dislocation, with periodic follow-up with an ophthalmologist.
- Careful monitoring of the skeletal system, especially during childhood and adolescence.
- Beta-blocker medications may be prescribed to lower blood pressure and, consequently, reduce stress on the aorta.
- Antibiotics may be prescribed prior to dental or genito-urinary procedures to reduce the risk of infection in people who experience mitral valve prolapse.
- Lifestyle adaptations, such as the avoidance of strenuous exercise and contact sports, are often necessary to reduce the risk of injury to the aorta.

What causes the Marfan syndrome?

A single abnormal gene causes the Marfan syndrome. Usually, this gene is inherited from a parent who is also affected. Approximately one-quarter of the cases occur as a result of a spontaneous mutation.

The Marfan syndrome is autosomal dominant, indicating that someone with the condition has a 50-50 chance that any offspring will inherit it.



Legislation

Since 1990, the NMF has testified annually before the House and Senate through the Coalition of Heritable Disorders of Connective Tissue. This enables the NMF to increase awareness in the legislative bodies about connective tissue disorders in general, and Marfan syndrome specifically, and encourage funding of research at the National Institutes of Health on these disorders.

Local Chapters

Nearly 75 chapters, support groups and phone contacts for people with Marfan syndrome have emerged. In addition to promoting the interests of the national organization, they also provide:

- A forum for affected people and their families to share experiences, provide support, and help alleviate the sense of isolation that often accompanies the diagnosis of Marfan syndrome.
- Local awareness, education and fundraising activities in which people can actively participate.
- An effective means for reaching out to local legislators to educate them about Marfan syndrome and encourage their support of funding for NIH research.

Research

The NMF established a Research Grant Program in 1986 to fund basic and clinical research on the Marfan syndrome. Since 1989, the NMF has awarded research grants to meritorious projects selected through a peer review by the NMF's Scientific Advisory Board. This program has become a catalyst for dramatic advances in Marfan syndrome research, including the identification of a chromosome and gene for the Marfan syndrome. The NMF has played a major role in coordinating efforts among scientific institutions and researchers worldwide, including:

- *Molecular Genetics and Cell Biology of the Marfan Syndrome Scientific Workshop*, Banbury Center of the Cold Spring Harbor (NY) Laboratories in 1991.

◆ *Second International Symposium on the Marfan Syndrome,* San Francisco, 1992, during which the discovery of the gene and chromosome that cause Marfan syndrome was shared with the international medical and scientific community for the first time.

◆ *Third International Symposium on the Marfan Syndrome,* Berlin, Germany, September 1994, which attracted more than 100 physicians and researchers from around the world.

◆ *Fourth International Symposium on the Marfan Syndrome,* Davos, Switzerland, scheduled for August 1996, which marks the 100th anniversary of the identification of the disorder by Dr. Antoine Marfan.

◆ *Marfan Syndrome and Related Disorder Genotype Consortium*, which serves as a clearinghouse for researchers worldwide to share information about new mutations in fibrillin-15, enabling the international scientific community to build upon each other's research rather than replicating it.

◆ *Marfan Syndrome and Related Disorders Clinical Database*, which serves as a resource for comparative statistics on research and treatment findings.

◆ *National Registry for Cardiovascular Surgery and the Marfan Syndrome*, which collects data on patients to determine the best surgical techniques for treatment of heart complications in the Marfan syndrome.

◆ *International Federation of Marfan Syndrome Organizations* (IFMSO), which provides standardized clinic and diagnostic criteria, and promotes the sharing of research findings worldwide. Marfan syndrome organizations from more than 20 countries are involved.

The NMF's national code for the
Combined Federal Campaign: #0547.

NMF 2/95

National Marfan Foundation

The National Marfan Foundation was founded in 1981 by people affected by Marfan syndrome and their families. Its purpose is to:

- Disseminate accurate and timely information about the condition to patients, family members and healthcare providers.
- Provide support for patients and their families.
- Support and foster research.

Programs

The National Marfan Foundation provides programs of information, awareness, education, service and research through a variety of projects.

Publications

- Pamphlets, booklets and other materials that explain the Marfan syndrome to both families and health care professionals are produced and distributed.
- A medical brochure series, targeted to health care professionals, addresses different medical disciplines involved with the Marfan syndrome.
- A quarterly newsletter, *Connective Issues*, provides regular communication to members about research, programs, new publications and local chapter news.
- An annual report provides an overview of the NMF's yearly activities and finances.

Annual Conference

The NMF sponsors a national conference each summer that includes speakers, panels and workshops. The meetings provide an opportunity to share information among affected people and medical professionals.

PLEASE RETURN THIS FORM TO:

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☐ Please send me additional information.
☐ I want to make a donation of $____.
☐ I would like to become a member of the NMF. My annual membership dues of $25 is enclosed.
☐ I cannot afford $25 dues, but please count me as a member of the NMF. I am able to donate $____ at this time.

I am interested in Marfan syndrome as a
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  ☐ Marfan syndrome suspected.
  ☐ Marfan syndrome diagnosis confirmed.
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A copy of the NMF's latest financial report may be obtained by writing to: New York State, Department of State, Office Charities Registration, Albany, NY 12231.

MARFAN SYNDROME

Physical Education and Activity Guidelines

Prepared by the
Professional Advisory Board

NMF
National Marfan Foundation

Exercise is important for people with the Marfan syndrome. It instills a sense of physical and psychological well-being. improves exercise endurance, lowers blood pressure, reduces weight, regulates metabolism and gastrointestinal function, increases bone density and physical strength, and often leads to beneficial lifestyle changes, such as smoking cessation, moderation of alcohol consumption and improved nutrition.

People with the Marfan syndrome and their families, as well as physical educators and healthcare professionals charged with overseeing the physical activity of people with the Marfan syndrome, should be aware of the following:

- Changes in connective tissue that give rise to the medical problems of the Marfan syndrome.
- Different forms of exercise and their impact on the Marfan syndrome.
- Basic guidelines for physical activities that should enable people with the Marfan syndrome to participate safely.

What are the changes in connective tissue that cause the Marfan syndrome?

The Marfan syndrome is a genetic disorder that affects the body's connective tissue. Connective tissue is found throughout the body and has various functions; it serves as the glue and scaffolding of all cells, giving structure and shape to organs, muscles, blood vessels and, in turn, the entire body.

One of the many ingredients of connective tissue is a protein known as *fibrillin*. In the Marfan syndrome, a diversity of problems occur in fibrillin, all caused by genetic mutation. As a result, the structure it provides to the body is weaker than normal. Because of this inherent weakness, some modifications in exercise are required in affected individuals. There is varia-

tion in the weakness of tissue among individuals, particularly in the wall of the aorta, the ligaments, the joints and the eyes; therefore, recommendations can be tailored to some degree.

What are the differences among forms of exercise and competition?

Exercise can be classified by a number of characteristics. *Aerobic* activities are conducted at an intensity that permits oxygen to be used to generate energy. There is a balance between the needs of the muscles and the ability of the body to provide oxygen to the muscles. If you can carry on a conversation while you are exercising, you are at an aerobic level.

In an *anaerobic* activity, there is insufficient oxygen, and cells have to rely on internal sources, which become depleted quickly, leading to fatigue. Anaerobic activity is usually of higher intensity, and is thus more stressful to tissues and the cardiovascular system.

When a muscle contracts through much of its full range of motion, such as the arm muscles when throwing a ball and the leg muscles when running, the exercise is called *isokinetic*. When the muscle is contracting without moving, such as when straining to lift a heavy weight or pushing a heavy piece of furniture, the exercise is called *isometric*. An increase in blood pressure, which stresses the heart and aorta, is greater with isometric exercise.

Most exercises and athletic activities involve combinations of isokinetic and isometric muscle work and aerobic and anaerobic energy use. The proportion of work and energy is determined by the nature of the activity, how strenuously a person is participating and, in team sports, even the position being played.

Sports are classified based on the risk of collision (contact) and on how strenuous they are. The following table shows one classification scheme.

Classification of Sports & Activities*

Before you apply this chart to your specific situation, it is important to realize that many sports can fall within several categories, depending on the intensity of your participation. It is essential to talk to your doctor about the sports and activities that are safe for you, and how to monitor your exertion level so that exercise remains safe throughout your involvement.

Contact/ collision high potential: strenuous	basketball field hockey ice hockey martial arts skiing (water) wrestling	boxing football lacrosse rodeo soccer
Limited contact: strenuous	baseball bicycling (intense) gymnastics horseback riding skating (ice & roller) skiing (downhill & cross-country) softball squash volleyball	
Noncontact: strenuous	aerobic dancing (high impact) crew running (fast) weightlifting	
Noncontact: moderately strenuous	aerobic dancing (low impact) badminton bicycling (leisurely) jogging swimming (leisurely) table tennis tennis	
Noncontact: nonstrenuous	golf riflery	bowling walking

* Modified from a classification devised by the American Academy of Pediatrics.

People with the Marfan syndrome should always avoid contact sports because of the risk of damaging the aorta and injuring the eyes. Strenuous activities also should be avoided because of the stress placed on the aorta.

Every activity has gradations, and no recommendation holds in all circumstances. For example, shooting baskets in the driveway is different from playing a full-court basketball game, and bicycling 10 miles in one hour on a level course is different from competing in a triathlon. To maximize safety of low intensity, non-contact activities, it is important to take necessary precautions, such as not carrying a heavy bag of golf clubs, and to avoid intense competitive efforts.

In short, it is essential for each individual with the Marfan syndrome to discuss physical activities, and specific activity levels, with his or her physician(s) so that exercise can be incorporated safely into the regular healthcare routine.

What if a person is taking medication?

Before beginning or increasing any exercise program, it is important to assess your current level of physical fitness, your health and your medications. The advice offered here is general, and is not meant to substitute for the recommendations of your personal physician.

Many people with the Marfan syndrome take a beta-blocker medication to reduce stress on the aorta. This medication lowers the pulse at rest and with exercise, and makes it somewhat more difficult to achieve a given level of physical fitness for the amount of physical work performed. Beta-blockers do permit a person to improve their endurance and strength while protecting the aorta. They do not, however, allow a person to perform very strenuous exercises or to play contact sports.

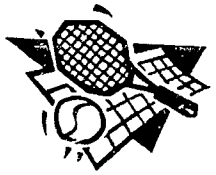
People who have artificial heart valves usually take an anticoagulant medication, Coumadin®. This medication interferes with blood clotting and increases the chances of bruising and internal hemorrhages. People taking Coumadin should avoid contact sports and any activity with a moderate risk of a blow to the head or abdomen. High-quality helmets should always be worn while bicycling.

What are some guidelines and modifications that permit safer exercise for people with the Marfan syndrome?

Physical activity modifications for people with the Marfan syndrome include the following:

◆ Favor non-competitive, isokinetic activity performed at a non-strenuous aerobic pace. Especially suited are sports in which you are free to rest whenever you feel tired and in which there is a minimal chance of sudden stops, rapid changes in direction, or contact with other players, equipment or the ground. Some beneficial activities are brisk walking, leisurely bicycling, slow jogging, shooting baskets, slow-paced tennis, and use of 1-3 pound hand weights.

◆ Choose an activity you enjoy that you can perform three or four times per week for 20-30 minutes. If time is a major constraint, three 10-minute sessions are nearly as effective as one 30-minute session.



◆ Stay at an aerobic level of work (about 50% of capacity). If you are on a beta-blocker or verapamil, keep your pulse under 100 beats per minute. If you are not on a beta-blocker, keep your pulse at less than 110. *Tip: It is often easier to feel the pulse over arteries in the neck than at the wrist.*

◆ Take your time and choose your activities wisely. With everyday activities, ask for help, make several trips carrying parcels rather than loading yourself down, use your legs rather than your back to lift, exhale when lifting, and refrain from straining to do anything.

◆ Avoid activities that involve isometric work, such as weightlifting, climbing steep inclines and pull-ups. If you are using a stationary cycle or a step-climber, keep the tension low. Multiple repetitions with a low resistance or weight are better than a few repetitions with a larger weight.

◆ **Do not test your limits.** This is particularly difficult for children during physical fitness tests in school and for people who once were competitive athletes. Be sure your child with the Marfan syndrome has an adaptive physical education program in place.

◆ Avoid activities that risk rapid changes in atmospheric pressure, such as scuba diving and flying in unpressurized aircraft. People with the Marfan syndrome are prone to collapse of a lung.

Exercise is beneficial to both the physical and emotional well-being of people with the Marfan syndrome. The average life expectancy of people with the disorder is now nearly 70 years, making regular gentle exercise an important general health measure. Most people with the Marfan syndrome should exercise regularly through low-intensity, low-impact activities adapted to meet their specific orthopedic, cardiovascular and ophthalmologic requirements. Always consult your personal physician before beginning any exercise program or performing any activity that may be strenuous for you.

NMF NationalMarfanFoundation

The *National***Marfan***Foundation* is a voluntary, tax-exempt foundation founded in 1981 by individuals affected by the Marfan syndrome and their families. The purpose of the NMF is to:

◆ Disseminate accurate and timely information about the disorder to patients, family members and the health care community.

◆ Provide means for patients and relatives to share experiences, support one another and improve their medical care.

◆ Support and foster research.

NMF activities are supported by membership dues. Please join us to help make a difference. As a member, you'll receive a quarterly newsletter, annual research updates and conference discounts. If you'd like to support our efforts and need information on becoming a member, please write to us at the address below.

Additional information (including videotapes) about the Marfan syndrome is available from the NMF.

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