National Marfan Foundation • 1981–2011

WHAT IS MARFAN SYNDROME? HOW MANY PEOPLE ARE AFFECTED/ IS IT ALWAYS INHERITED?

Marfan syndrome is a genetic disorder that weakens multiple body systems, including the heart, blood vessels, bones and joints, lungs and eyes. The life-threatening part of Marfan syndrome is the weakening of the aorta, the large blood vessel that carries blood away from the heart. Early diagnosis and treatment are essential for maximizing life expectancy. It is estimated that more than 200,000 people in the U.S. are affected by the Marfan syndrome or a related disorder. Experts agree that about half of those affected with Marfan syndrome are not diagnosed. Marfan syndrome is often hereditary, but 25 percent of affected people are the first in their family to have the disorder.

HOW LONG CAN PEOPLE WITH MARFAN SYNDROME LIVE? WHY IS IT SO IMPORTANT FOR PEOPLE TO GET ANY EARLY DIAGNOSIS?

There is no cure for Marfan syndrome, but with an early diagnosis, proper treatment and careful management of the disorder, the life span can be extended into the 70's. Without the diagnosis and treatment, people with the disorder are at up to 250 times increased risk of a tear or rupture of the aorta, which can be fatal.

WHAT ARE THE CHARACTERISTICS OF MARFAN SYNDROME?

Because connective tissue makes up the entire body, the disorder manifests itself in many body systems. The most common characteristics are:

- Tall stature - affected people are usually, but not always, taller than other people in their family
- Arms, legs, fingers and toes are disproportionately long, as compared to the trunk
- Loose-jointedness
- Indented or protruding chest bone
- Scoliosis
- Flat feet
- Nearsightedness
- Dislocated lens

WHAT IS THE MOST DANGEROUS PART OF MARFAN SYNDROME?

Aortic enlargement. Without medications and lifestyle modifications (e.g., no competitive or contact sports), the aorta is prone to enlarge and could dissect (tear) or rupture. An aortic rupture is usually fatal.

HOW IS MARFAN SYNDROME TREATED?

People affected by Marfan syndrome should be treated by a physician familiar with the condition and how it affects all body systems. Careful management includes:

- Annual echocardiogram to monitor the size and function of the heart and aorta.
- An initial eye exam, including a slit-lamp exam, by an ophthalmologist, with periodic follow up exams.
- Careful monitoring of the skeletal system, especially during childhood and adolescence, by an orthopedist.
- Medications may be prescribed to lower blood pressure and, consequently, reduce stress on the aorta.
- Lifestyle adaptations to reduce stress on the aorta.

In addition to routine monitoring, people with the condition should visit specialists to manage and treat the body systems that are affected.

WHERE CAN PEOPLE GET MORE INFORMATION ABOUT THE DISORDER?

The National Marfan Foundation is the only dedicated resource for complete, up-to-date, accurate information on Marfan syndrome and related disorders. For additional information about Marfan syndrome, please contact the National Marfan Foundation, 800-8-MARFAN (862-7326), or staff@marfan.org.

THE NATIONAL MARFAN FOUNDATION

The National Marfan Foundation is a non-profit voluntary health organization dedicated to saving lives and improving the quality of life of individuals and families affected by Marfan syndrome and related disorders by:

- Educating affected individuals, family members and the health care community
- Advocating for and funding basic and clinical research
- Providing a network of local and special-interest support groups to help affected people and their families

# # #