# Marfan's Syndrome

Marfan's syndrome is an inherited disorder resulting from an abnormality of the connective tissue, which helps to hold the various tissues of the body together. In Marfan's syndrome, the connective tissue abnormality leads mainly to problems in the skeleton (bones), heart and blood vessels, and eyes. There is no cure, but regular medical care can reduce the rate of serious complications.

## What is Marfan's syndrome?

Marfan's syndrome is a genetic (inherited) condition affecting 1 in 5000 to 1 in 10,000 newborns. Your child is born with this disease, even if it isn't recognized until later. Marfan's syndrome is caused by an abnormal gene affecting the body's connective tissue, which works like glue to help hold various tissues together.

Children with Marfan's syndrome may have abnormalities of the bones and joints, heart and blood vessels, eyes, and skin. The high risk of heart problems is the main concern. Children with Marfan's syndrome don't all have the same problems or disease severity. Your child will receive close medical attention to keep him or her as healthy as possible and to prevent complications.

## What kinds of problems occur with Marfan's syndrome?

- The typical person with Marfan's syndrome is tall and thin, with long legs, arms, and fingers. The face may also appear long and thin.
- Heart and blood vessel diseases are the most serious complications of Marfan's syndrome. Because of the connective tissue abnormality, the aorta (the main artery through which blood is pumped out of the heart) may become widened and stretched (dilated). This may lead to weakening and bulging of the aorta ("aneurysm") and tearing of its walls ("dissection"). Leaks and other problems of the heart valves can also develop.
- Abnormalities of the skeleton may include a "caved-in" chest (called pectus deformity), abnormal curve of the spine (called scoliosis), and crowding of the teeth. Your child may have loose joints ("double-jointed") that are easily dislocated.
- Various eye problems may occur, including dislocation of the lens of the eye, nearsightedness (myopia), glaucoma, cataracts, and detached retina.
- Loose skin, sometimes with stretch marks.
- Lung problems: as your child ages, he or she will be at increased risk of a problem called "pneumothorax." In this condition, the lungs leak air, putting pressure on them and causing chest pain and difficulty breathing.
- Your child may not have all of these abnormalities. Even when caused by the same genetic abnormality, the disease is more severe in some patients than in others.

## What causes Marfan's syndrome?

- An abnormality of the fibrillin gene. Fibrillin is essential for development of connective tissue and is found in nearly every organ of the body.
- Most often, the abnormal gene is inherited from a parent. If one parent has an abnormal fibrillin gene, each child will have a 50% chance of inheriting Marfan's syndrome. Genetic counseling can help you understand your chances of passing on the disease to future children.
- In 25% of people with Marfan's syndrome, the gene mutation (which means a change in genetic material occurred) that made the abnormal fibrillin gene happened when your child was conceived; neither parent has an abnormal gene. In this situation, other children in your family are not necessarily at increased risk of Marfan's syndrome.

## How is Marfan's syndrome diagnosed?

- There is no specific test for Marfan's syndrome. The diagnosis is based on the presence of typical abnormalities, including heart and eye problems, and information on your family's medical history. Genetic testing may be possible.
- Once Marfan's syndrome is diagnosed, tests will be performed to check the various organs and body systems:
  - A test called an echocardiogram ("echo"), which uses sound waves to take pictures of the heart.
  - A slit-lamp examination (a test using a special light to examine the inside of the eye).
  - Other tests may be recommended as well.

## How is Marfan's syndrome managed?

Various doctors may be involved in your child's care: for example, a cardiologist (heart specialist), an ophthalmologist (eye doctor), a medical geneticist (specialist in genetic diseases), and an orthopedic surgeon (bone and joint specialist). Our office will help to coordinate your child's care.

Regular, repeated evaluations of all of the organ systems involved are essential to protecting your child's health and to detecting any complications as early as possible:

- *Heart problems.* A key concern is to monitor enlargement of the body's central main artery (the aorta) and the functioning of the heart valves. The cardiologist will determine what tests and possible treatment are needed. Your child may have to follow certain precautions, including:
  - Avoiding overexertion. Gentler forms of exercise, such as bicycling and swimming, are a good choice.
  - Wearing a Medic-Alert bracelet or necklace to alert health care providers that he or she has Marfan's syndrome.
  - If your child has heart valve problems, he or she may need to take antibiotics before dental procedures or surgery. This is done to prevent a serious condition called infective endocarditis (infection of the heart valves and tissues lining the heart).
- *Bone and joint problems.* Scoliosis (abnormal spinal curvature) and other bone and joint problems are possible. The orthopedic surgeon will evaluate and treat these problems.
- *Eye problems.* We will probably recommend visits to an eye doctor (ophthalmologist) for evaluation of possible eye problems. Yearly eye examinations are essential for your child.
- *Pregnancy.* Sexually active women and girls with Marfan's syndrome must be especially careful about birth control. Although it is possible for women with this disease to have successful pregnancies, the risks are a lot higher than for women without it.

Family or individual counseling may help your child and family deal with the stresses of living with Marfan's syndrome.

## When should I call your office?

Your child with Marfan's syndrome will be scheduled for regular medical visits, including screening for heart disease and other problems.

Between visits, call our office or go the emergency room if your child develops:

- Signs of problems related to the heart or lungs, such as chest pain or shortness of breath.
- Eye problems—any change in vision.

## Where can I get more information about Marfan's syndrome?

The National Marfan Foundation on the Internet at *www.marfan.org*, or call 1-800-8-MARFAN (1-800-862-7326).

Please type your custom instructions and/or office contact information here.