



## BASIC INFORMATION

### DESCRIPTION

A rare, inherited connective tissue disorder. Connective tissue supports and adds strength to other body tissue and body parts such as tendons and ligaments and heart valves. The severity of the symptoms varies greatly among patients. It affects males and females equally.

### FREQUENT SIGNS AND SYMPTOMS

- Tall and thin body shape.
- Long arms and legs.
- Long, thin fingers (arachnodactyly).
- Chest deformity (may be sunken in or protrude out).
- Curved spine (scoliosis).
- High palate in the mouth and overcrowded teeth.
- "Double jointed;" joint weakness or looseness.
- Dislocation of eye lens, usually upward.
- Myopia (nearsighted).
- Symptoms of heart problems (eg., shortness of breath, tiredness, irregular or rapid heart beat).
- Stretch marks on the shoulders, hips and lower back.
- Easy bruising or bleeding (uncommon).

### CAUSES

Inherited disorder in about 85% of cases. Other cases occur with no known cause. The disorder is present from birth and diagnosis can sometimes be made in newborns. However, signs or symptoms are often not seen until adolescence or young adulthood.

### RISK INCREASES WITH

- Advanced paternal age may be a risk in those cases that are not clearly inherited.
- Family history of Marfan syndrome.

### PREVENTIVE MEASURES

- No preventive measures. Prenatal diagnosis is possible in some cases.
- Each child has a 50% chance of inheriting the disorder from an affected parent. Children may be more or less severely affected.
- Get genetic counseling if you have Marfan syndrome or there is a family history of the disorder.

### EXPECTED OUTCOMES

Early diagnosis and steps to prevent and treat complications have helped to improve the outlook for patients. Lifespan is about the same as an average person.

### POSSIBLE COMPLICATIONS

- Cardiovascular problems can be life-threatening.
- Bacterial endocarditis (heart inflammation).
- Complications of the aorta (heart artery).
- Heart and lung problems (due to chest deformity).
- Depression and anxiety.
- Retinal detachment (rare).



## DIAGNOSIS & TREATMENT

### GENERAL MEASURES

- Your health care provider will do a physical exam and ask questions about family medical history. There is no one test to diagnose Marfan syndrome. Medical tests include echocardiogram (for diagnosing heart problems) and an eye exam. Genetic testing may be done.
- Treatment will involve a team approach that involves eye, cardiac, orthopedic, and dental care. Drugs are often used to help prevent complications.
- Annual screening echocardiograms are recommended beginning in adolescence in order to detect the start of any complications. X-rays of spine are needed during growth years to detect scoliosis.
- Have annual eye exams. Special lenses and eye drops or surgery may be needed for lens dislocation.
- Surgery may be needed for cardiovascular problems.
- Scoliosis may require use of a brace or surgery.
- Surgery may be needed if chest bones are sunken in.
- Pregnant women with Marfan syndrome are managed as high-risk patients. Outcome is usually excellent.
- To learn more: National Marfan Foundation, 22 Manhasset Ave., Port Washington, NY 11050; (800) 862-7326; website: [www.marfan.org](http://www.marfan.org).

### MEDICATIONS

- Beta-blockers are often prescribed for heart problems.
- Hormone therapy may be given prior to puberty.
- Antibiotic therapy may be prescribed.

### ACTIVITY

- Fully active unless limited by symptoms. Exercise helps physical and emotional well-being. Get medical advice before starting any exercise or sports activity.
- You may be advised to avoid contact sports, activities that risk head injury, rapid decompression (scuba diving), isometric exercises (weightlifting, pull-ups, etc.).

### DIET

No special diet.



## NOTIFY OUR OFFICE IF

- You believe your child has signs or symptoms of Marfan syndrome.
- After diagnosis, symptoms change or worsen.

Special notes:

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More notes on the back of this page