Marfan syndrome is a disorder that affects connective tissue. Connective tissue supports many parts of your body. You can think of it as a type of “glue” between cells.

Marfan syndrome can affect many parts of the body, such as:

- Skeleton.
- Heart and blood vessels.
- Eyes.
- Skin.
- Nervous system.
- Lungs.

It is usually passed from parent to child through the genes.
Marfan syndrome is passed down from your parents. You have a 50 percent of getting the disease if one of your parents has it. Men, women, and children can have Marfan syndrome. It is found in people of all races and ethnic backgrounds.

What are the symptoms?

Marfan syndrome affects people in different ways. Some people have only mild symptoms, and others have severe problems. Most of the time, the symptoms get worse as the person gets older. It affects many parts of the body, including the:

- **Skeleton.** People with Marfan syndrome are often very tall, thin, and loose jointed. They may have:
  - Bones in the arms, legs, fingers, and toes that are longer than normal.
  - A long, narrow face.
  - Crowded teeth because the roof of the mouth is arched.
  - A breastbone that sticks out or caves in.
  - A curved backbone.
  - Flat feet.

- **Heart and blood vessels.** Most people with Marfan syndrome have problems with the heart and blood vessels, such as:
  - A weak part in the vessel that carries blood from the heart. This can make the blood vessel tear or break.
  - Heart valves that leak, causing a “heart murmur.” Large leaks may cause:
    - Shortness of breath.
    - Tiredness.
    - Very fast or uneven heart rate.

- **Eyes.** Some people with Marfan syndrome have problems with the eyes, such as:
  - Nearsightedness.
  - Glaucoma (high pressure within the eye) at a young age.
  - Cataracts (the eye’s lens becomes cloudy).
  - A shift in one or both lenses of the eye.
  - A detached retina.

- **Skin.** Many people with Marfan syndrome have:
  - Stretch marks on the skin. These are not a health problem.
  - A hernia (part of an organ that pushes out of its space).

- **Nervous system.** When people with Marfan get older, the tissue surrounding the brain and spinal cord may weaken and stretch. This affects the bones in the lower spine. Symptoms of this problem include:
  - Pain in the stomach area.
  - Painful, numb, or weak legs.

- **Lungs.** People with Marfan syndrome do not usually have problems with their lungs. If there are symptoms, they include:
  - Stiff air sacs in the lungs.
  - A collapsed lung if the air sacs become stretched or swollen.
Snoring or not breathing for short periods while sleeping.

What causes it?

Connective tissue is made of many kinds of protein. Marfan syndrome is caused by a problem with a gene that makes one of these proteins.

Is there a test for it?

There is no single test for Marfan syndrome. Your doctor may use many tools to see if you have the disease:

- Medical history (whether you have had any symptoms).
- Family history (any family members who have Marfan syndrome or who died at a young age from heart problems).
- A physical exam, including the length of the bones in the arms and legs.
- An eye exam.
- Heart tests.

How is it treated?

There is no cure for Marfan syndrome, but treatment can help. You should see your doctor on a regular basis to treat or even prevent some problems.

- **Skeleton.**
  - Get a yearly exam of the spine and breastbone.
  - Use a back brace.
  - Have surgery for serious back problems.

- **Heart and blood vessels.**
  - Get regular checkups and heart tests.
  - See a doctor or go to an emergency room for pain in the chest, back, or stomach area.
  - Wear a medical alert bracelet.
  - Take medicine for heart valve problems.
  - Have surgery to replace a valve or repair the blood vessel from the heart if the problem is severe.

- **Eyes.**
  - Get yearly eye exams.
  - Wear eyeglasses or contact lenses.
  - Have surgery if needed.

- **Lungs.**
- Do not smoke (because it can hurt your lungs).
- See a doctor if you have any problems breathing during sleep.
- **Nervous system:** Take medicine for back pain.

**Pregnancy.** Women with Marfan syndrome can have healthy babies. But pregnancy is high risk, since it stresses the heart. If you are planning to become pregnant, talk to your doctor about whether you should have surgery to reduce this risk. If you are already pregnant, see your doctor right away to prevent problems with your heart.

**Who treats it?**

You may need special kinds of doctors to treat Marfan syndrome. Your health care team may include:

- Family doctor or pediatrician.
- Cardiologist, a doctor who treats heart problems.
- Orthopaedist, a doctor who treats bone problems.
- Ophthalmologist, a doctor who treats eye problems.
- Geneticist, a doctor who specializes in genetic diseases.

**Living With It**

Advances in medicine now make it possible for people with Marfan syndrome to live about as long as the average person. However, the disease can cause strong emotions, such as anger and fear. You may also be worried that you children will have the disease. Children with Marfan syndrome might find it hard that they can’t play some sports.

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Ways to help live with the disease include:

- Get proper medical care and correct information.
- Find strong social support.
- Eat a balanced diet and maintain a healthy lifestyle.
- Get medium levels of exercise to keep the skeleton and heart healthy. You should not play contact or competitive sports. You should also not do exercises where you tighten the muscles without moving them (“planks” are an example).
- Get genetic counseling, which may help you learn about the disease and the risk of passing it on to your children.
For More Info

U.S. Food and Drug Administration
Toll free: 888-INFO-FDA (888-463-6332)
Website: https://www.fda.gov

Drugs@FDA at https://www.accessdata.fda.gov/scripts/cder/daf. Drugs@FDA is a searchable catalog of FDA-approved drug products.

Centers for Disease Control and Prevention, National Center for Health Statistics
Website: https://www.cdc.gov/nchs

National Heart, Lung, and Blood Institute
Website: https://www.nhlbi.nih.gov

National Human Genome Research Institute
Website: https://www.genome.gov

Marfan Foundation
Website: http://www.marfan.org

National Organization for Rare Disorders
Website: https://www.rarediseases.org

American Heart Association
Website: https://www.americanheart.org

March of Dimes Birth Defects Foundation
Website: http://www.marchofdimes.com

If you need more information about available resources in your language or other languages, please visit our webpages below or contact the NIAMS Information Clearinghouse at NIAMSInfo@mail.nih.gov.

- Asian Language Health Information
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