Points To Remember About Marfan Syndrome

- Marfan syndrome is a genetic disorder that leads to problems with the development of connective tissue in the body.
- Changes to a specific gene cause Marfan syndrome, and most people inherit the disorder from their parents.
- Treatment depends on the area of the body affected and may include medications, other treatments, and surgery to manage the condition and its complications.
- You can do several things to manage the disorder, such as see your doctors regularly; reach out for support from loved ones, other people with the disorder, and mental health professionals; eat a healthy, well-balanced diet; and avoid smoking.

What is Marfan syndrome?

Marfan syndrome is a genetic disorder that affects the body’s ability to make healthy connective tissue, which supports the bones, muscles, organs, and tissues in your body. The condition can affect different areas of the body, including:

- Bones, ligaments, tendons, and cartilage.
- Organs, such as the heart and lungs.
- Skin.

Who gets Marfan syndrome?

Men, women, and children can have Marfan syndrome. It is found in people of all races and ethnic backgrounds.

What are the symptoms of Marfan syndrome?

The symptoms of Marfan syndrome vary from person to person because connective tissue is found throughout the body. Some people have mild or just a few symptoms, while others can have more serious problems. Symptoms may include:

- Abdominal pain.
• Arms, legs, fingers, and toes that are much longer in relation to the rest of the body.
• Chest that caves in or sticks out.
• Curved spine.
• Flat feet.
• Headaches.
• Heartbeat that feels like it is skipping a beat, fluttering, or beating too hard or too fast.
• Highly arched mouth that can lead to crowding of teeth.
• Loose joints.
• Long, narrow face.
• Low back pain and numbness in the legs.
• Shortness of breath due to changes in the lungs or heart.
• Stretch marks on the skin.
• Vision changes, such as blurry vision and extreme nearsightedness.

What causes Marfan syndrome?
A mutation or change in a gene causes Marfan syndrome. Most people inherit the changed gene from their parents, but some children are born with the syndrome, even though there is no family history of the disorder. This is caused by a new change in the gene.

Is there a test for Marfan syndrome?
No single test can diagnose Marfan syndrome. To see if you have the disorder, your doctor may:
• Ask about your family and medical history.
• Perform a physical exam.
• Order blood and imaging tests.
• Order genetic testing to look for the changed gene that causes Marfan syndrome.

How is Marfan syndrome treated?
Although there is no cure for Marfan syndrome, doctors use treatments to relieve symptoms and prevent additional problems or complications. Treatment depends on the area of the body affected by the syndrome and may include:
• Medications to help manage pain and problems with your heart.
• Other treatments, such as braces.
• Surgery to help correct problems with your bones or eyes.
Who treats Marfan syndrome?

Health care providers who may treat Marfan syndrome include:

- Cardiologists, who treat issues with the heart.
- Cardiothoracic surgeons, who specialize in surgery of the heart, lungs, and other organs and tissues in the chest.
- Clinical geneticists, who diagnose and treat children and adults with genetic disorders.
- Dental providers, such as orthodontists and oral-maxillofacial surgeons.
- Dermatologists, who specialize in conditions of the skin, hair, and nails.
- Mental health professionals, who provide counseling and treat mental health disorders, such as depression and anxiety.
- Nurse educators, who help you understand your condition and help start treatment plans.
- Occupational therapists, who teach how to safely perform activities of daily living.
- Ophthalmologists, who treat eye disorders.
- Orthopaedists, who treat bone problems.
- Pediatricians, who diagnose and treat children.
- Physical therapists, who teach ways to build muscle strength.
- Primary care doctors, such as family physicians or internal medicine specialists, who coordinate care between the different health providers and treat other problems as they arise.
- Pulmonologists, who treat lung problems.

Living with Marfan syndrome

Treating and living with Marfan syndrome, and its complications, is a lifelong process. However, treatment makes it possible for people with the disorder to have long, productive lives. The following tips can help you manage the disorder:

- Ask your doctors how often you should schedule follow-up visits. The visits may include regular eye exams, image tests to check for heart and lung problems, and an evaluation of your skeleton and growth.
- Reach out for support. Talk to your family and friends about the disorder and your feelings. Consider joining a community or online support group.
- Seek counseling or talk to a health care professional if you feel depressed or anxious about Marfan syndrome and its effects on your body.
- Eat a healthy well-balanced diet rich in fruits, vegetables, and whole grains.
- Consider wearing medical identification to notify providers about your condition during an emergency.
- If you smoke, quit. Smoking can have a negative impact on both your bone and lung health.
Women with Marfan syndrome can have healthy pregnancies. However, the pregnancy is high risk because it can add stress on the heart. If you are thinking about getting pregnant, talk to your doctor. Planning helps doctors treat problems prior to the pregnancy to keep both the mother and baby healthy.

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

U.S. National Library of Medicine, Genetics Home Reference
Website: https://ghr.nlm.nih.gov/condition/marfan-syndrome#diagnosis

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

March of Dimes
https://www.marchofdimes.com

Marfan Foundation
https://www.marfan.org

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

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
- Spanish Language Health Information