

What is Marfan Syndrome?

Marfan syndrome (MFS) is a common inherited disorder affecting the connective tissue that connects or helps support different structures in the body. MFS is reported to affect 1 in 3000 to 5000 individuals. The syndrome is associated with classic ocular, cardiovascular, and musculoskeletal abnormalities, although involvement of the lungs, skin, and central nervous system may also occur. Patients with MFS are also at high risk of experiencing aortic complications, including aortic root dilatation and dissection.

How is it Transmitted?

Marfan syndrome is an autosomal dominant condition caused by a mutation in the *FBN1* gene. This means that if a child receives a copy of the mutated gene, they will be affected by MFS. Also, a parent with MFS has a 50% chance of passing it on to their children. However, approximately 25% of patients develop the syndrome as a result of a new (spontaneous) mutation involving the *FBN1* gene. In less than 10% of patients with typical Marfan phenotype, no mutation in *FBN1* is identifiable.

How is it Diagnosed?

A diagnosis is usually established with a genetic test and the presence of either aortic root enlargement or ectopia lentis (displacement of the lens). For this, a defined set of criteria (the Ghent nosology) have been developed to facilitate the recognition of the syndrome and improve patient management and counseling. Since MFS can be inherited, those who have family members with MFS are more likely to contract it and should be screened regularly. Moreover, some family members may have a genetic mutation without knowing it; hence it is important to ask your medical professional if family screening is necessary.

Is There a Treatment for MFS?

There is no current treatment that cures MFS, but specific interventions may improve certain aspects of the syndrome. Because connective tissue is affected throughout the body, regular follow-up with different medical specialists is important. Ongoing care may include medication, lifestyle changes, imaging tests, and surgery when needed. Outcomes improve with early diagnosis, treatment to slow or prevent enlargement of the aorta, and timely elective surgery.

What Can I do Every Day?

- 1 Always carry an alert card and/or wear medical alert jewelry
- 2 Follow prescribed medical follow-up procedures, in collaboration with your attending physician.
- 3 Inform the people around you about warning signs, so that they can help you if necessary.
- 4 Let them know you have an alert card or medical alert jewelry.
- 5 Inform your doctor if you are planning to become pregnant or are in early pregnancy.

What Physical Activity Can I Practice?

Do: Participate in regular, light to moderate physical activity where you can speak easily without stopping to catch your breath. Examples include light weight lifting without straining, traditional yoga, light pilates, tai chi, walking, hiking, swimming, and cycling. Non-competitive sports with minimal sudden stops or rapid changes in direction, such as golf, bowling, leisure badminton, leisure doubles tennis, and leisure volleyball, are also acceptable. Always wear proper protective equipment.

Avoid: Vigorous exercise that causes muscle fatigue, straining, or exhaustion, participating in contact sports or sports with a high risk of shock, and participating in activities that risk rapid atmospheric pressure changes (changes in altitude).

Marfan Syndrome Manifestation & Recommendations

Body System & Manifestations

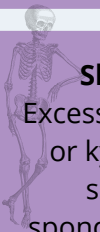
Treatment and Recommendations



Cardiovascular (Heart & Blood Vessels):

Aneurysm (bulging), dissection (tear), or rupture of the aorta or other arteries; mitral or tricuspid valve prolapse; cardiomyopathy (weak heart muscle)

Imaging: Echocardiography is used for diagnosis and to monitor the heart valves and aortic root. CT or MRI may be used for more detailed imaging of the heart and aorta. An electrocardiogram (ECG) may also be used to detect abnormal heart rhythms. After aortic root replacement surgery, CT or MRI is performed regularly to monitor aortic size. **Medication:** Beta-blockers and angiotensin receptor 2 blockers or ACE inhibitors may be used to slow aortic enlargement, lower blood pressure, and improve heart function. **Surgery:** Preventive or emergency surgery may be needed to repair aneurysms or heart valves. Pacemakers or defibrillators may be implanted if serious abnormal heart rhythms occur. Seek emergency care for sudden severe chest or back pain.



Skeletal (Bones):

Excessive growth; scoliosis or kyphosis (abnormal spine curvature); spondylolisthesis (slipped vertebra); pectus excavatum or carinatum (chest wall deformities)

Imaging: X-rays and MRI may be used to check spinal curves (such as scoliosis) and spinal stability, including the neck. **Treatment:** Braces may be used to support the spine. Surgery may be needed for severe scoliosis, chest wall deformities (pectus), or spinal instability. Hormone therapy may be used during puberty to help control rapid growth. **Lifestyle:** Avoid contact or high-impact sports and choose low-impact activities. Physiotherapy can help strengthen muscles and reduce joint or muscle pain. Occupational therapy may help adapt daily activities. Avoid spinal manipulation by a chiropractor.



Allergic/Inflammatory:

Allergies; gastrointestinal inflammatory disease; eosinophilic esophagitis

Medication/Surgery: In case of known allergies, carry an adrenaline pen on you, if advised. **Feeding tubes** help with caloric intake. Treatments may be prescribed by doctor. **Lifestyle:** Avoid specific foods, exposure to environmental allergens, and caffeine-rich foods and drinks. Eat fiber-rich foods to avoid constipation. Ensure a well-balanced diet and regular food intake. Avoid fluoroquinolone antibiotics.



Lungs:

Restrictive lung disease, pneumothorax (collapsed lung); asthma; sleep apnea (stopping and restarting of breathing during sleep), emphysema, interstitial disease

Imaging: Chest x-rays, CT, or MRI may be used to assess lung problems. Spirometry may be used to evaluate breathing and help identify conditions such as asthma or emphysema. Sleep studies may be done to diagnose sleep apnea. **Treatment:** Inhalers with bronchodilators and corticosteroids may be used to treat breathing symptoms. A breathing device may be used during sleep for sleep apnea. **Surgery:** A chest tube may be needed to treat a collapsed lung (pneumothorax). Seek emergency care for sudden chest pain or trouble breathing. Talk to your doctor if you have severe daytime fatigue.



Eyes:

Retinal detachment; ectopia lentis; severe myopia/astigmatism.

Treatment: Any sudden onset of visual disturbance is an indicator of a complication. Immediately call an ophthalmologist for a consultation. Glasses or eye drops may be prescribed to improve vision. Patients with MFS should be followed by an ophthalmologist for the detection and management of complications. In severe cases, surgical removal of the lens may be required.

Other potential manifestations: skin striae; recurrent or incisional hernia; high-arched palate; dural ectasia, intracranial hypotension

**This is not an exhaustive list of manifestations. MFS patients will not present with all of these manifestations, and having these manifestations does not mean a person has MFS.*

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