

Marfan Syndrome

A plain-English guide for you and your family

WHAT IS HAPPENING

Normally, your body makes a special protein called fibrillin-1 that acts like the steel cables in a bridge, holding connective tissues together and keeping them strong yet flexible. In Marfan syndrome, a change in one gene means the body makes fibrillin-1 that is weak or missing. Without good fibrillin-1, the walls of blood vessels (especially the main artery from the heart), the structures holding the lens in the eye, and the ligaments around bones stretch too much under everyday stress. Over time, the aorta can balloon out or tear, the lens can slip out of place, and the skeleton grows longer and more flexible than usual. With careful monitoring and treatment, people with Marfan syndrome can live full lives.

What you might feel

- Tall stature with long arms, legs, and fingers
- Chest pain or pressure
- Vision problems or lens dislocation
- Shortness of breath with exertion
- Pectus deformity (sunken or protruding breastbone)
- Joint hypermobility or frequent dislocations
- Sudden severe back or abdominal pain
- Skin stretch marks not related to weight change

Call the doctor right away if...

- Sudden severe chest or back pain (possible aortic dissection—activate code team and order STAT CT angiography)
- New heart murmur with fainting (syncope) or heart failure symptoms (acute valve rupture or severe regurgitation—urgent cardiology consult)
- Blood pressure difference >20 mmHg between arms (dissection extending into brachiocephalic vessels—immediate imaging)

- Shortness of breath with unilateral absent breath sounds (spontaneous pneumothorax—needs chest X-ray and possible chest tube)

What the care team will do

- Monitor for aortic dissection (tear in the large blood vessel leaving the heart) and rupture
- Assess cardiovascular status including heart sounds and murmurs
- Monitor for spontaneous pneumothorax (collapsed lung)
- Screen for vision changes and lens dislocation
- Educate about activity restrictions and avoidance of contact sports
- Administer beta-blockers as prescribed and monitor heart rate and blood pressure
- Provide genetic counseling education and family screening referrals
- Assess for scoliosis (curved spine) and joint hypermobility complications

Medicines you may be given — and why

Atenolol

Atenolol blocks beta-1 receptors on heart cells so adrenaline (epinephrine) cannot bind and speed up the heart. Slower heart rate and weaker contractions lower blood pressure and reduce the force hitting the aortic wall with each heartbeat. Less mechanical stress on the aorta slows aneurysm growth.

Losartan

Losartan blocks angiotensin II from binding to AT1 receptors on blood vessels and the heart. This lowers blood pressure and also reduces TGF- β (a signaling protein that damages aortic tissue in Marfan syndrome). Less TGF- β means less breakdown of elastin (stretchy fibers) and collagen in the aortic wall.

Warfarin

Warfarin blocks vitamin K epoxide reductase, the enzyme that recycles vitamin K in the liver. Without active vitamin K, the liver cannot make clotting factors II, VII, IX, and X. Thinner blood prevents clots from forming on mechanical heart valves (which all Marfan patients with valve replacements may have).

Celiprolol

Celiprolol blocks beta-1 receptors to slow the heart and lower blood pressure, like atenolol. It also mildly stimulates beta-2 receptors in blood vessels, causing vasodilation (widening). This

combination may reduce aortic stiffness more than pure beta-blockers. Some European studies show benefit in vascular Ehlers-Danlos, and it is sometimes used off-label in Marfan.

Pneumococcal and influenza vaccines

These vaccines train the immune system to recognize *Streptococcus pneumoniae* and influenza virus. After vaccination, B cells produce antibodies that bind to bacterial capsule or viral surface proteins. If the patient later encounters the real pathogen, preformed antibodies quickly neutralize it, preventing pneumonia. Marfan patients with chest-wall deformities or prior pneumothorax have reduced lung reserve, so preventing respiratory infections is critical.

Genetic counseling and prenatal diagnosis

Genetic counselors explain the 50% autosomal dominant inheritance risk, review family pedigree, and offer preimplantation genetic testing (PGT) or prenatal diagnosis. PGT involves in-vitro fertilization, embryo biopsy at 5 days, and FBN1 gene sequencing before uterine transfer. Alternatively, chorionic villus sampling (CVS) at 10–13 weeks or amniocentesis at 15–20 weeks can test fetal DNA. These methods allow parents to make informed reproductive choices.

Where this information comes from

- American Heart Association (AHA) 2022 Guidelines for Management of Patients with Thoracic Aortic Disease
- American College of Medical Genetics and Genomics (ACMG) 2021 Practice Guidelines for Genetic Evaluation of Marfan Syndrome
- National Marfan Foundation 2023 Clinical Guidelines for Diagnosis and Management
- European Society of Cardiology (ESC) 2020 Guidelines on the Diagnosis and Treatment of Aortic Diseases

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