

Marfan Syndrome

Marfan syndrome is a genetic condition where the body makes faulty connective tissue. The main protein that gives strength and elasticity to blood vessels, eyes, and bones does not work properly. This causes dangerous stretching of the aorta, lens problems in the eyes, and tall stature with long limbs.

THE ONE-LINE TRUTH

Marfan syndrome is a connective tissue disorder caused by defective fibrillin-1 protein, leading to fragile elastic fibers in the aorta, eyes, and skeleton that stretch and tear under normal body stress.

The chain — how it unfolds

1. Defective fibrillin-1 causes progressive aortic root dilation — why: Each heartbeat stretches the weak aortic wall slightly more, and the vessel loses the ability to recoil, so diameter increases gradually over years
2. Aortic regurgitation develops as the root dilates — why: When the aortic root widens, the three valve leaflets cannot meet properly in the center, allowing blood to leak backward into the left ventricle during diastole (relaxation phase)
3. Left ventricular volume overload and eccentric hypertrophy — why: The ventricle must pump both the regurgitant volume and the normal stroke volume, so it dilates and thickens to handle the extra load
4. Increased wall stress accelerates aortic dilation — why: According to Laplace's law, wall tension equals pressure times radius, so as the aorta gets wider, wall stress increases exponentially, creating a vicious cycle of worsening dilation
5. Aortic dissection occurs when intimal tear allows blood into the vessel wall — why: Extreme wall stress causes the inner aortic layer (intima) to tear, and high-pressure blood cleaves through the weakened media layer, separating wall layers
6. Blood flow to branch vessels is compromised by the dissection flap — why: The false lumen (dissection space) can compress the true lumen or occlude branch vessel origins, cutting off blood to the brain, kidneys, intestines, or limbs

7. Cardiac tamponade (the heart squeezed by fluid around it) develops if dissection ruptures into the pericardium — why: Blood fills the pericardial sac around the heart, preventing ventricular filling and causing cardiogenic shock
8. Death from aortic rupture, tamponade, or end-organ malperfusion — why: Sudden massive bleeding (hemorrhage) into the chest or abdomen causes exsanguination (bleeding to death), or critical organs fail from lack of oxygen delivery

Spot it — what you see, and why

- Tall stature with long arms, legs, and fingers: Defective fibrillin-1 protein (a connective tissue building block) allows bones to grow longer than usual because growth plates stay open longer and cartilage is less restrained
- Chest pain or pressure: Weakened aortic wall stretches and tears, or mitral valve leaflets prolapse backward into the left atrium causing starved blood flow (ischemia) or dissection
- Vision problems or lens dislocation: Zonules (tiny fibers made of fibrillin that hold the lens in place) stretch or break, letting the lens move out of position
- Shortness of breath with exertion: Mitral or aortic valve regurgitation (backward leak) forces the heart to pump the same blood twice, or spontaneous pneumothorax (lung collapse) reduces oxygen exchange
- Pectus deformity (sunken or protruding breastbone): Weak costal cartilage (rib connective tissue) and rib overgrowth allow the sternum to cave inward or push outward
- Joint hypermobility or frequent dislocations: Ligaments (bands that hold bones together) have abnormal fibrillin so they overstretch and fail to stabilize joints

Red flags — act now

- 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)

Treat it — in this order

1. Beta-blocker therapy (atenolol or metoprolol) → Slows heart rate and reduces aortic wall stress (force per unit area) by lowering blood pressure and the speed of ventricular contraction, slowing aortic root growth by approximately 0.1 cm/year
2. Preventive (Prophylactic) aortic root replacement surgery → Replaces the dilated ascending aorta and aortic valve with a graft (tube) and mechanical or bioprosthetic valve, preventing rupture or dissection; 30-year survival exceeds 80% post-surgery
3. Angiotensin II receptor blocker (ARB) therapy (losartan) → Blocks TGF- β (transforming growth factor beta) signaling that drives aortic wall remodeling and smooth muscle loss, reducing aortic root growth rate in some studies by 0.1–0.2 cm/year
4. Competitive and contact sports restriction → Avoids sudden blood pressure spikes and chest trauma that can trigger dissection or worsen valve regurgitation; dynamic low-intensity exercise (walking, swimming) is safe and improves fitness
5. Mitral or aortic valve repair or replacement → Restores competent valve closure, reduces left ventricular volume overload, and prevents progression to heart failure or pulmonary high blood pressure (hypertension); mitral repair has lower long-term complications than replacement

Your top priorities at the bedside

1. Monitor for aortic dissection (tear in the large blood vessel leaving the heart) and rupture — why: Marfan syndrome weakens the aortic wall, making it prone to life-threatening tears. Early detection through regular blood pressure checks and assessment of chest or back pain can save lives.
2. Assess cardiovascular status including heart sounds and murmurs — why: Mitral valve prolapse (backward bulging of a heart valve) and aortic regurgitation (leaky aortic valve) are common in Marfan syndrome and can lead to heart failure if not monitored and treated.
3. Monitor for spontaneous pneumothorax (collapsed lung) — why: Patients with Marfan syndrome have weak connective tissue in the lungs, increasing risk for sudden lung collapse. Watch for sudden chest pain, shortness of breath, or decreased oxygen levels.