Marfan Syndrome

George Silvay | Joshua A. Heller

Risk

- Prevalence is estimated at 1:5000 people.
- Inherited as autosomal dominant trait, though 25% of cases are sporadic.
- The 2010 Ghent Nosology for Marfan Syndrome guides Dx, heavily prioritizing aortic root dilation, ectopia lentis, and family history.

Perioperative Risks

- Aortic rupture and dissection, mitral valve prolapse, mitral or aortic valve regurgitation, arrhythmias, pneumothorax, restrictive lung disease, and chest wall and spine deformity

Worry About

- Ascending aortic dissection and rupture
- Mitral and aortic valvular insufficiency
- Myocardial ischemia due to medial necrosis of coronary arteries
- Dyspnea, reduced functional residual capacity increased risk of pneumothorax

Overview

- Connective tissue disorder typically inherited via autosomal dominant genetics. Pathophysiology is complex but characterized by a defect of collagen synthesis, which decreases tensile strength and elasticity of connective tissue. CV, particularly aortic, manifestations are most responsible for reduced life expectancy, but the disorder has pansystemic implications.
- CV manifestations are most lethal. Pts commonly diagnosed and monitored via transthoracic echocardiography.
- Invasive management recommended for type A dissection, type B dissection with severe pain, ischemia, rapid aortic growth, or large aortic diameter. Prophylactic surgery recommended when aortic root exceeds 50 mm or 46 mm in setting of adverse family history, severe valvular involvement, rapid aortic dilation, or planned pregnancy.
- Ocular manifestation that is most defining is ectopic lentis, which is the subluxation of the lens
- Pts may have a high-arched palate, crowded teeth, abnormal skull shape, malar hypoplasia, or retrognathia. Pts are at risk for spontaneous pneumothorax, restrictive lung disease, and obstructive sleep apnea.
- Musculoskeletal features include increased length of long bones, joint laxity, scoliosis, pectus excavatum and carinatum, laxity of cervical spine, and lumbar dural ectasia.

Etiology

- Mutation in *FBNI*, the gene on chromosome 15 that encodes fibrillin-1, is an extracellular matrix glycoprotein. Fibrillin-1 normally binds TGFβ. Decreased functional fibrillin-1 leads to excess TGFβ, which is thought to lead to a cascade of inflammatory degradation of elastic fibers and extracellular matrix.
- The 2010 Revised Ghent Nosology defines diagnostic criteria for Marfan syndrome, including data from aortic imaging, genetic testing, family history, and physical examination.

Usual Treatment

- Pts are prescribed beta-blocker therapy and potentially ACE-inhibitor or ARB therapy to reduce aortic wall stress and potentially growth rate. The American Heart Association has recommendations for physical activity limitations because straining and Valsalva actions can worsen aortic wall stress.
- Regular follow-up by a cardiologist is necessary to monitor heart and aorta; and elective aortic root, aorta, and valve repair common to avoid catastrophic complications.

Assessment Points

System	Effect	Assessment by Hx	PE	Test
HEENT	Ectopia lentis	Myopia	Retinal detachment, lens subluxation	Ophthalmoscopy
CV	Aortic dissection Myocardial ischemia	Chest pain Angina		MRI, ECHO, CT ECG Stress testing Angiography
	Arrhythmias	Palpitations	Heart rate Chest auscultation	Holter monitor Electrophysiology
RESP	Restrictive lung disease Obstructive sleep apnea	Dyspnea Snoring Decreased functional capacity	Scoliosis Abnormal airway exam	Pulm function testing CXR Sleep study
MS	Tall stature Joint hypermobility Recurrent joint dislocation Hernias TMJ dysfunction		Chest wall deformity Elevated arm to height ratio	

Key References: Radke RM, Baumgartner H: Diagnosis and treatment of Marfan syndrome: an update, *Heart* 100(17):1382–1391, 2014; Castellano JM, Silvay G, Castillo JG: Marfan syndrome: clinical, surgical, and anesthetic considerations, *Semin Cardiothorac Vasc Anesth* 18(3):260–271, 2013.

Perioperative Implications

Preoperative Preparation

- Antibiotics for SBE prophylaxis if a prosthetic valve or other indication from 2007 AHA Guidelines.
- Consider periop beta-blockade to mitigate increases in aortic wall tension.
- Consider large-bore IV access and transfusion capacity in case of vascular rupture if vascular manipulation possible.

Monitoring

- Standard ASA monitoring, and consider transesophageal echocardiography and CNS monitoring (such as cerebral oximetry)
- Invasive monitoring as appropriate for planned surgery

Airway

- High-arched palate, crowded teeth, and retrognathia are characteristic.
- Potential cervical laxity and instability with extension.
- Potential for TMJ dislocation with direct laryngoscopy.
- If a known aortic aneurysm, consider fiber optic bronchoscopy to inspect for compression of respiratory tract.

Preinduction/Induction

- Meticulous hemodynamic control to avoid increases in aortic wall tension
- Careful positioning to avoid dislocations

Maintenance

- No specific technique is known to be superior. Hemodynamic vigilance is critical.

Extubation

- Avoid sudden swings in hemodynamics because this may increase aortic wall tension and sheer forces.
- Be cognizant of risk of myocardial ischemia.

Adjuvants

- Adequate pain management is important.
- Pts may have different dose response to neuraxial medication due to lumbar dural ectasia, which may have significance for dosing and distribution of local anesthetic.

Anticipated Problems/Concerns

- CV: Aortic dissection, mitral or aortic regurgitation, myocardial ischemia, and cardiac arrhythmias
- Respiratory: Difficult airway, high pneumothorax risk, and restrictive lung disease with thoracic deformity