orphanaesthesia

Anesthesia recommendations for

Marfan Syndrome

Disease name: Marfan Syndrome

ICD 10: Q87.4

ORPHAcode: 558

Synonyms: Marfan's syndrome

Disease summary: Marfan syndrome is an autosomal dominant, multisystem disease with a reported incidence of 1 in 3,000 to 5,000 individuals [1]. There is a broad range of clinical severity associated with Marfan syndrome, ranging from isolated features to neonatal presentation of severe and rapidly progressive disease. Classic manifestations involve ocular (lens dislocation, myopia), cardiovascular (aortic root dilation with aortic regurgitation, mitral valve prolapse with mitral regurgitation), and musculoskeletal abnormalities (long bone overgrowth, scoliosis, kyphosis, joint hypermobility), however, involvement of the lung (pneumothorax), skin (striae), and central nervous system (dural ectasia) is also common in Marfan syndrome [2]. The modified Ghent criteria for diagnosis incorporate genetic testing, the systemic score, ectopia lentis, and family history [3]. Marfan syndrome is caused by autosomal dominant mutations in the FBN1 gene located on chromosome 15, which encodes the extracellular matrix protein fibrillin-1 responsible for classic Marfan syndrome. However, up to 30% of cases occur without an affected parent and represent de novo mutations [4]. Prophylactic treatment with β-blockers is considered standard care in adults (unless contraindicated) and has been shown to reduce the rate of aortic dilation. There is no definitive recommendation for either general (GA) or regional anesthesia (RA). Regardless of anesthetic technique, care should be taken to prevent a sudden increase in myocardial contractility, producing an increase in aortic wall tension, which could lead to aortic dissection [5].

Diagnosis may be incorrect; if uncertainty exists, the diagnosis should be re-evaluated.

Every patient is unique; individual circumstances must always guide clinical care.

Medicine is in progress; new clinical knowledge may not be yet reflected in this recommendation.

Recommendations are not rules or laws; they provide a framework to support clinical decision-making. Although this recommendation has passed a structured review process, it does not meet the formal criteria of a guideline.

Translations may not always reflect the most recent updates of the English version.

Find more information on the disease, its centers of reference and patient organizations on Orphanet: www.orpha.net

A	**AIRWAY / ANESTHETIC TECHNIQUE**	Airway management challenges: – High-arched palate – Micrognathia – Temporomandibular joint laxity – Generally normal cervical spine mobility; use normal precautions during neck extension (atlantoaxial instability is very rare in Marfan) GA considerations: – Risk of aortic dissection with hypertensive surges – Avoid excessive sympathetic stimulation (e.g., during laryngoscopy) – Requires careful hemodynamic control RA considerations: – Spinal or epidural techniques may be complicated by dural ectasia (seen in up to ≈90% of patients) – May result in unpredictable spread or failed block
B	**BLOOD PRODUCTS (COAGULATION)**	– Special preparation or storage of blood products is not routinely required – No primary coagulopathy is typical of Marfan syndrome – If aortic aneurysm/dissection repair or major cardiovascular surgery is planned: • Full crossmatch and coagulation studies preoperatively are prudent • Bleeding risk is increased due to the nature of cardiovascular surgery, not because of an intrinsic coagulation disorder
C	**CIRCULATION**	No congenital heart disease, but cardiac pathology is common, including: – Aortic root dilation – Aortic aneurysm – Risk of aortic dissection (especially with hypertension) – Mitral valve prolapse (MVP) and mitral regurgitation – Arrhythmias (often atrial) may occur in patients with cardiac involvement.

		– Increased risk of heart failure, particularly in those with aortic insufficiency or prior cardiac surgery – Anesthetic goals: avoid hypertension, tachycardia, and large swings in preload/afterload
D	**DRUGS**	Avoid sympathomimetic agents that elevate BP and stress the aorta (e.g., ketamine). Intraoperative BP/HR control: short-acting β-blockers (e.g., esmolol) or vasodilators as needed. Patients may already be on β-blockers or angiotensin receptor blockers; requires careful hemodynamic monitoring. Anxiolysis is important to prevent BP surges; consider midazolam or clonidine for controlled preoperative BP/HR. No known association between Marfan syndrome and MH or rhabdomyolysis.
E	**EQUIPMENT**	Careful BP monitoring required; consider invasive arterial line for high-risk or major surgery. Intraoperative TEE may be useful for cardiac/aortic assessment. Postoperative ICU monitoring may be indicated after major surgery or in patients with significant cardiac history.

Typical surgery and procedures

The literature review is limited to case reports/case series of patients presenting for surgical treatment of scoliosis, retinal detachment, orthodontic surgery, cesarean delivery, aortic root replacement, elective repair of thoracic aortic aneurysm, and emergency repair of aortic dissection. Patients with Marfan syndrome have increased incidence of inguinal, femoral and umbilical hernia, recurrent pneumothoraces, requiring surgical treatment, as well as arthropathies, severe pectus deformities, necessitating orthopedic correction. Bentall and De Bono were pioneers in the "Bentall" procedure to replace the dilated aortic root in 1968, improving patients' life expectancy [6].

Type of anesthesia

There is no definitive recommendation for either GA or RA. A distinct advantage of GA is that, if acute dissection should occur, the airway is protected and immediate cardiac surgery is possible. Disadvantages of GA include the risk of hypertensive response to intubation, which could predispose to aortic dissection, if not prevented pharmacologically. There are no contraindications for sedation in these patients. However, in patients with significant pulmonary disease (emphysema, restrictive lung disease due to scoliosis) this technique should be considered with caution [7].

Necessary additional preoperative testing (beside standard care)

Cardiovascular complications are recognized to be the major cause of morbidity and mortality in patients with this disease. Patients develop aneurysms involving the aortic root (sinuses of Valsalva) and are at risk for aortic dissection. Descending aortic and abdominal aortic aneurysms (AAAs) are less common. Type B aortic dissection is the initial aortic event in about 10% of patients and may also occur despite previous root replacement [5]. Transthoracic echocardiography (TTE) or computed tomography (CT) or cardiac magnetic resonance imaging (MRI) of thoracic aorta (if the aortic root, ascending aorta, or both are not adequately visualized on TTE) should be ordered preoperatively. Left ventricular dilation may predispose to alterations of repolarization and fatal ventricular arrhythmias and could be identified on ECG [8].

Patients with significant dilation of the aortic root should be referred to a cardiothoracic surgeon for evaluation of aortic root replacement before elective surgery. There have been case reports of cesarean delivery followed by repair of aortic dissection in parturients, therefore women with aortic dilation >4 cm, measured at the level of Valsalva sinuses, or history of aortic dissection should deliver in a center where cardiovascular surgery is available [9].

The 2022 guidelines of the American College of Cardiology (ACC), American Heart Association (AHA), and American Association for Thoracic Surgery (AATS) recommend an elective operation for patients with Marfan syndrome at an external diameter of ≥ 5 cm to avoid acute dissection or rupture. Indications for repair at an external diameter < 5 cm include rapid growth (>3 mm/y), family history of aortic dissection at a diameter less than 5 cm, desire for pregnancy and presence of progressive aortic regurgitation [5]. Prophylaxis against infective endocarditis in the presence of valvular abnormality is not required unless the patient has a mechanical valve.

Patients with severe scoliosis should undergo pulmonary function testing to evaluate the extent of restrictive lung disease [5]. Due to the high prevalence (70% incidence in the lumbosacral area) of dural ectasia (increased diameter of the dural sac) in this patient population, ideally MRI of the spine should be ordered if possible before planning any neuraxial procedure, even in the absence of the symptoms (low back pain, headache, proximal leg pain, weakness and numbness above and below the knee, and genital/rectal pain) [10].

In patients suffering from Marfan syndrome, caudal epidural anesthesia should only be performed after a radiological diagnostic examination. The absence of symptoms does not exclude meningeal abnormalities [11].

Particular preparation for airway management

Preoperative examination of the airway is critical because the presence of micrognathism and high arched palate with crowded teeth may make visualization of the larynx during direct laryngoscopy difficult [3]. Clinically symptomatic cases of atlantoaxial dislocation in these patients are rarely reported in the literature and screening radiographs of the cervical spine for patients with Marfan syndrome undergoing GA are not routinely recommended [11,12,13].

If a large aortic aneurysm is present, consider an awake fiber-optic evaluation of the airway to assess any compression or deviation of airway structures before securing the airway [14]. There is one case-report of tracheomalacia in a patient with Marfan syndrome manifested by increased airway pressures and difficulty with ventilation during anesthesia in a prone position [15]. Obstructive sleep apnea has been reported in patients with Marfan syndrome, although

there are no case reports describing difficulties managing the airway due to this comorbidity [16].

Patients are at increased risk of developing pneumothorax, which should be kept in mind during positive pressure ventilation [3]. Although there is a possibility of temporomandibular joint dysfunction, this condition has not been reported to cause difficulty with laryngoscopy. Excessive traction on laryngoscopy should be avoided to prevent temporomandibular joint dislocation [17]. Cardiovascular response during laryngoscopy should be blunted pharmacologically. The hemodynamic goal is to reduce the stress imposed upon the wall of the aneurysm [5].

Particular preparation for transfusion or administration of blood products

There is no evidence from the literature that patients with Marfan syndrome have preexisting coagulation abnormalities associated with a higher risk of blood transfusion. Patients requiring anticoagulation due to prosthetic valve or aortic root should be offered a bridging therapy before elective surgery [3].

Particular preparation for anticoagulation

There is no information suggesting the need for particular anticoagulation, except in patients following valve or aortic root replacement.

Particular precautions for positioning, transportation and mobilization

Patients must be carefully positioned and handled on the operating table and stretchers to avoid joint dislocations and injuries secondary to joint laxity [7].

Interactions of chronic disease and anesthesia medications

Patients may be taking β-blockers, angiotensin receptor blockers (ARB) for blood pressure (BP) control to minimize shear forces and wall stress in the aorta, diuretics (history of heart failure), and anticoagulants, if they had valve or aortic root replacement in the past. β-blockers should be continued perioperatively [18].

One of the populations that could potentially benefit the most from treatment with β-blockers is the pediatric population, and the rationale is that treatment with β-blockers may allow surgery to be delayed and, therefore, the eventual implantation of a larger graft, which may, in turn, avoid the need for re-intervention at a later time. Potential adverse effects of perioperative β-blockade include bradycardia and hypotension. Continuing ARBs up to the time of surgery increases perioperative hypotension. Omitting diuretics on the morning of surgery minimizes hypovolemia and electrolyte disturbance [5].

Anesthetic procedure

Patients with Marfan syndrome require special considerations regarding the anesthetic technique to avoid extreme hypotension and hypertension, conserve coronary perfusion, and prevent increases in aortic wall tension and thus prevent dissecting the aneurysm. Labetalol and nitroglycerin should be available to treat hypertensive episodes, and hemodynamically stable induction should be performed [19]. This may be achieved using short-acting opioids such as remifentanil administered via a target-controlled infusion regimen. Volatile anesthetics have the potential to decrease the force of cardiac ejection, therefore decreasing the risk of aortic dissection. Phenylephrine is the vasopressor of choice, because ephedrine may induce tachycardia via its β-adrenergic effect. Avoidance of excessive endogenous catecholamine production through control of pain and anxiety is essential [5].

Volatile agents or neuromuscular blocking agents are generally avoided if somatosensory evoked potentials or motor evoked potentials monitoring is necessary. In this case, anesthesia will rely on a propofol infusion (50-100 µg/kg/min) to provide anesthetic maintenance.

Hypothermic circulatory arrest (HCA) for the repair of the adult aortic arch has become a standard technique in thoracic aortic surgery. But a prolonged deep HCA greater than 30 minutes is associated with the occurrence of severe neurological damage [20]. Three major neuroprotective techniques in HCA for the repair of the adult aortic arch have been championed in the contemporary era: profound hypothermia alone, retrograde cerebral perfusion, and ante-grade cerebral perfusion. To ensure optimal anesthesia management, close communication between the anesthesia team, surgical team, perfusionist, operating room nurses, and staff in the intensive care unit is necessary.

Antibiotic prophylaxis: According to the latest AHA guidelines, patients with isolated valvular abnormality do not require antibiotic prophylaxis against infective endocarditis. If the patient has a history of a prosthetic cardiac valve or a history of infective endocarditis in the past, preoperative antibiotic prophylaxis should be administered according to AHA guidelines [21].

Antibiotic prophylaxis is recommended for all dental procedures that involve manipulation of gingival tissues or periapical region of teeth or perforation of the oral mucosa, procedures on respiratory tract or infected skin, skin structures, or musculoskeletal tissue. Antibiotic prophylaxis solely to prevent IE is not recommended for GU or GI tract procedures, as well as is not recommended for vaginal deliveries [21].

Particular or additional monitoring

In addition to standard American Society of Anesthesiologists' (ASA) monitoring, an arterial line should be used intra-operatively to monitor for sudden changes in the BP. The use of intraoperative transesophageal echocardiography (TEE) to monitor aortic root diameter has been reported in several case reports [19]. The use of central venous access in patients with Marfan syndrome is not dictated by the diagnosis itself, but rather by standard clinical indications for central line insertion.

Possible complications

Patients with Marfan syndrome and left ventricular dilation are at risk of ventricular arrhythmias. Aortic root diameter greater than 4 cm carries a risk of aortic dissection. Stanford Type A

dissection is the commonest type seen in patients with Marfan syndrome. Spontaneous coronary artery dissection has also been reported in the literature [8].

Postoperative care

The degree of postoperative monitoring depends on the surgical procedure and the preoperative condition of the patient, particularly the size of the aortic root and left ventricle. Not all Marfan patients require ICU care postoperatively, though it should be considered for higher-risk cases [22].

Disease-related acute problems and effect on anesthesia and recovery

Due to the associated high mortality, anesthetic management of patients with severe aortic root dilation is a challenging situation. Elastic fiber degeneration, lacking smooth muscle cells in aorta and mucopolysaccharide deposition in between the cells of the media are the main features of aorta in Marfan syndrome [7].

Aortic dissection in patients with Marfan syndrome can be precipitated by major hemodynamic changes under anesthesia. Hemodynamic instability can be minimized with adequate monitoring and preparation during intubation, intraoperative fluid resuscitation, planned extubation, and adequate analgesia. These goals can prevent this life-threatening event [19].

Ambulatory anesthesia

There are no reported experiences in patients with Marfan syndrome in the ambulatory anesthesia setting. The author's opinion is that low risk, minimally invasive surgery could be performed on patients with Marfan syndrome in the ambulatory setting, in the absence of significant involvement of cardiovascular and respiratory systems.

Obstetrical anesthesia

All women with genetically proven Marfan syndrome should have counseling on the risk of dissection and the recurrence risk and have a complete multidisciplinary evaluation including imaging of the entire aorta before pregnancy. Pregnancy should be discouraged in women with previous aortic dissection because of the high risk for aortic complications [5].

Dissection occurs most often in the last trimester of pregnancy (50%) or the early postpartum period (33%) [23,24]. In all women with known aortic disease and/or an enlarged aortic root diameter, the risks of pregnancy should be discussed before conception. Aortic root diameter >4 cm during pregnancy carries a higher risk of dissection (10% compared to 1% in patients with aortic root diameter <4 cm) and conception is not advised if the root diameter is >4.5 cm. Depending on the aortic diameter, patients with aortic pathology should be monitored by echocardiography at 4-12-week intervals throughout their pregnancy and 6 months postpartum because aortic root enlargement may be accelerated by pregnancy. Pregnancy should be supervised by a cardiologist and obstetrician who are alert to the possible complications [25]. Instrumented vaginal delivery can be safely performed in patients with Marfan syndrome who have no cardiovascular involvement or stable minimal aortic dilation (<4 cm). Epidural

analgesia is strongly recommended to minimize the stress associated with labor pain. Cesarean delivery is indicated in patients with an aortic diameter >4.5 cm, aortic dissection, severe aortic regurgitation or heart failure. Cesarean delivery should also be considered in the presence of contraindications for epidural analgesia for vaginal delivery or if epidural analgesia has failed, to avoid the stress on the dilated aortic root, caused by untreated labor pain. For patients with aortic root diameter 4.0-4.5 cm, the decision about the method of delivery should be individualized and involve a multidisciplinary team approach, including an obstetrician, anesthesiologist and cardiologist. Family history of dissection, rapid growth during pregnancy should be taken into account. It is essential to continue β-blocker therapy during pregnancy, peripartum and postpartum period to prevent aortic dissection.

Parturients with Marfan syndrome will only require anticoagulation if they have a mechanical valve [26].

Both RA and GA have been used successfully in parturients undergoing cesarean delivery [27]. Neuraxial anesthesia may pose technical challenges due to kyphoscoliosis. The standard dose of local anesthetic required for the spinal anesthesia might be inadequate due to the presence of dural ectasia, therefore combined spinal-epidural anesthesia might be a technique of choice for cesarean delivery [28]. Local anesthetic without epinephrine is preferable for epidural anesthesia in case of accidental intravascular injection. Although dural ectasia is not an absolute contraindication for epidural anesthesia, the theoretical risk of increased incidence of dural puncture or inadequate anesthesia should be discussed with the patient [29]. Structural changes in the ligamentum flavum have been suggested as a likely cause of accidental dural puncture. Ultrasound examination of the back before placing an epidural catheter may decrease the risk of puncture of the enlarged dural sac. Ascending and descending aortic dissection has been reported in the postpartum period in parturients with Marfan syndrome, therefore symptomatic women should undergo immediate investigations [30]. Asymptomatic patients still need to undergo an echocardiographic examination before hospital discharge.

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