Marfan's Syndrome

Autosomal dominant disorder caused by a defect in the gene encoding fibrillin which polymerizes and combines with other proteins to become a microfibril; involves skeleton, eye, and CV system, skin, fascia, lungs, skeletal muscle, CNS, and adipose tissue.

ANESTHETIC CONSIDERATIONS:

- Airway risk
  - TMJ laxity and prone to dislocation with laryngoscopy
  - C1 / C2 disease, potential for dislocation
- Respiratory insufficiency
  - RLD due to scoliosis and pectus carinatum / excavatum
  - Spontaneous pneumothorax risk
- Significant risk of CVS disease and perioperative complications
  - Aortic aneurysm and dissection – need for prevention of HTN / increased CO
  - MI – need for ischemia monitoring and risk assessment perioperatively
  - MVP, MR, AR
  - Arrhythmias & conduction defects

ANESTHETIC GOALS:

- Avoid sudden increases in aortic wall tension
- Hemodynamic goals for MR and MVP
- Restrictive pattern ventilation
- Anticipate risk of pneumothorax
- Careful positioning witih attention to joints and peripheral nerves
- Caution with airway manipulation: TMJ and C1 / C2 disease

HISTORY

- Airway - TMJ dislocation with laryngoscopy
- Respiratory - RLD due to scoliosis, pectus carinatum or excavatum; bronchogenic cysts; spontaneous pneumothorax
- CVS - Aortic disease (cystic medial necrosis leading to dissection), aortic aneurysms, PA dilatation
  - Coronary artery abnormalities (medial necrosis of arterioles)
  - MVP, MR, AR
  - Arrhythmias and conduction disturbances (coronary insufficiency involving vascular supply of SA & AV nodes)
- Neurological - Lens dislocation, myopia, retinal detachment
- MSK - Tall stature, joint hypermobility, recurrent dislocation, hernias

PHYSICAL

- **HEENT** – airway exam, TMJ exam
- **CVS** – standard exam including pulses for rhythm
- **RESP** – standard exam and pectus scoliosis (RLD)

INVESTIGATIONS

- **Labs**
  - ABGs
- **Imaging**
  - ECG, ECHO, TEE, MIBI
  - MRI or ECHO (dilatation of aortic root)
  - CT chest, angiography
- **Special**
  - PFTs

OPTIMIZATION

- Consider antibiotics for SBE prophylaxis
- Consider perioperative beta-blockade treatment to mitigate increases in myocardial contractility and aortic wall tension (dP/dT)

ANESTHETIC OPTIONS

- Local, regional GA acceptable
- Regional may be difficult if significant scoliosis
  - May require ↑ doses of local anesthetic due to ↑ size and enlargement of the neural canal

ANESTHETIC SETUP

- **Drugs**
  - Standard
- **Equipment**
  - Routine monitors along with 5-lead ECG (ST-segment analysis)
  - TEE, and invasive monitors as indicated (care with probes and catheters)

MANAGEMENT OF ANESTHESIA

- **Induction**
  - Airway → remember high arched palate, potential cervical laxity / instability
    - Careful laryngoscopy to avoid TMJ dislocation

- Avoid HTN → prevent sudden ↑ in contractility or LV ejection velocity
- Cautious PPV → gentle PPV as patients prone to pneumothorax
- Careful positioning to avoid dislocations

- **Maintenance**
  - Carefully monitor for myocardial ischemia
  - Ensure patient has deep anesthetic as to avoid ↑ CO or significant HTN
  - No one technique has shown superiority
- **Emergence**
  - Avoid sudden increases in CO, BP as this may increase dP/dT
  - High risk for developing myocardial ischemia

DISPOSITION & MONITORING

- Postoperative observation for CVS complications, fast a.fib, pneumothorax, retinal detachment
- Adequate pain management important

COMPLICATIONS

- CV: aortic dissection, MVP, MR, AR, MI, cardiac arrhythmias
- Respiratory: pneumothorax, restrictive lung disease with thoracic deformity

PATHOPHYSIOLOGY

- Autosomal dominant
- Defect in collagen synthesis resulting in ↓ tensile strength and elasticity of CT
- ↑ Long bone length
- Arachnodactily, joint laxity, ↓ muscle tone, scant SQ fat, high arched palate, pectus carinatum or excavatum and pes palnus
- Arm span > height
- 95% of deaths due to CVS complications

REFERENCES

- Roizen & Fleisher – Essence of Anesthesia Practice – p215