Ehlers Danlos Syndrome

A group of inherited connective tissue disorders (9 distinct subtypes) due to abnormal production of procollagen and collagen.

ANESTHETIC CONSIDERATIONS:

- Life threatening cardiovascular manifestations (Type IV)
  - Aortic aneurysm
  - Spontaneous rupture of medium and large vessel arteries
  - Valvular disease: Mitral regurgitation
  - Cardiac conduction abnormalities (rare)
- Increased risk of spontaneous pneumothorax
- Coagulopathy and increased risk of bleeding
- Positioning considerations:
  - Joint hypermobility and skin fragility
  - Association with kyphoscoliosis

ANESTHETIC GOALS:

- Careful positioning
- Minimize airway trauma due to increased risk of bleeding
  - Avoid nasopharyngeal/esophageal manipulation
- Minimize airway pressures during PPV
  - Increased risk of spontaneous pneumothorax
- Avoid regional and neuraxial techniques due to increased risk of bleeding
- Hemodynamic goals for mitral regurgitation (if present)
- Minimize swings in blood pressure to avoid increases in transmural pressure and prevent aneurysm rupture or dissection.

HISTORY

- Muscle weakness
- Skin fragility and hyperelasticity
- Easy bruising
- Joint hypermobility
- Osteoarthritis
- GI: spontaneous bowel rupture
- GU: uterine rupture

PHYSICAL

- **AIRWAY:** tracheal dilation, TMJ hypermobility
- **CNS**
- **CVS:** mitral regurgitation, cardiac conduction abnormalities, medium and large vessel spontaneous rupture, aortic aneurysm
- **RESP:** spontaneous pneumothorax
- **GI:** increased risk of bowel perforation
- **GU:** increased risk of uterine rupture, preterm labor
- **HEME:** easy bruising, extensive ecchymosis with minimal trauma
- **ENDO**
- **MSK:** joint hypermobility, ecchymosis, poor wound healing, skin fragility

INVESTIGATIONS

- **LABS:** CBCcd (bleeding risk), lytes, BUN, Cr, INR, PTT
- **ECG:** cardiac conduction abnormalities
- **ECHO:** assessment of mitral valve
- **CT:** evaluate aneurismal disease

OPTIMIZATION

ANESTHETIC OPTIONS

- GA
- Regional: increased risk of hematoma formation
- Neuraxial: increased risk of hematoma formation

ANESTHETIC SETUP

- **Standard Machine setup**
- **Standard emergency drugs**

MANAGEMENT OF ANESTHESIA

COEXISTING PAGE 444:

"There is no specific recommendations for the selection of drugs to provide anesthesia. Regional anesthesia is not recommended because of the tendency of these patients to bleed and form extensive hematomas."

- **Induction**
- **Maintenance**
- **Emergence**

DISPOSITION & MONITORING

COMPLICATIONS

- **Bleeding**
- **Postoperative wound dehiscence**
- **Spontaneous pneumothorax**
- **Vascular rupture**

OBSTETRICS

- Increased risk of preterm labor, premature rupture of membranes and uterine rupture (due to abundance of Type III collagen in uterine smooth muscle)
- Neuraxial analgesia relatively contraindicated

PATHOPHYSIOLOGY

- Group of inherited connective tissue disorders due to abnormal procollagen and collagen production
- Prevalence: 1:5000
- At least 9 different subtypes:
    - Type I (46%):
        - Autosomal Dominant
        - severe disease with marked skin laxity and scarring
        - severe joint hypermobility and increased risk of dislocations
        - poor wound healing
        - kyphoscoliosis
        - hallux valgus, flat feet
        - hematoma formation less common
        - mitral valve prolapse
        - Obstetrics: premature rupture of membranes.
    - Type II (35%)
        - Milder form
        - Autosomal dominant
        - Same features as type I
        - Less skin fragility and bruising
        - Usually digits are the only joints involved
    - Type III (10%)
        - Autosomal dominant
        - Benign familiar hypermobile form
        - Minimal to no skin changes
        - Severe joint hypermobility with increased risk of osteoarthritis
    - **Type IV (6%)**
        - Arterial form; decreased or absent type III collagen synthesis
        - Autosomal recessive or autosomal dominant inheritance
        - White translucent skin with vessels easily visible underneath
        - Skin is fragile but not hyperextensible
        - Multiple bruises, scars, keloids, molluscoid pseudotumors
        - Joint hyperextensibility is rare or absent
        - INCREASED RISK OF SUDDEN DEATH
            - Mitral valve prolapse and regurgitation
            - Spontaneous pneumothorax
            - Arterial aneurysms and spontaneous vascular rupture
        - Decreased life expectancy
        - Low weight, short stature
    - Type V (5%)
        - X linked recessive
        - Highly extensible skin with orthopedic abnormalities
        - Bruising and joint hyperextensibility is rare
    - Type VI (2%)
        - Ocular form; also somewhat dangerous with severe disease
        - Autosomal recessive
        - Severe disease: bruising, extensible skin, poor wound healing, pigmented scarring
        - Joint hyperextensibility
        - Ocular involvment: retinal hemorrhage, retinal detachment, glaucoma, scleral coloration, globe rupture (rare)
    - Type VII (3%)
        - Arthrochalasis multiplex congenita (AR or AD)
        - Joint hyperextensibility with less severe skin changes
        - Spontaneous joint dislocation
        - Short stature
    - Type VIII
        - Periodontal form
        - Rare
        - Autosomal dominant
        - Gingival inflammation
        - Gingival resorption and loss of teeth by age 30
        - Joint and skin involvement is variable
    - Type IX
        - X linked cutis laxa
        - X linked recessive
        - Bony prominence on occiput
        - Chronic diarrhea and orthostatic hypotension
        - Poor wound healing

REFERENCES

- Stoeltings Anesthesia and Coexisting Disease Chapter 18
- Emedicine “Ehlers Danlos Syndrome”