Mucopolysaccharidoses

The mucopolysaccharidoses are genetically determined diseases in which mucopolysaccharides are stored in tissues in abnormal quantities and excreted in large amounts in the urine. The disorders result from a deficiency of a specific lysosomal enzyme that is required to break down these compounds. As a result, mucopolysaccharides accumulate in tissues, producing specific clinical manifestations.

ANESTHETIC CONSIDERATIONS:

- Difficult airway – Difficult BMV and difficult laryngoscopy visualization
- C-spine instability
- Systemic involvement:
    a. Restrictive respiratory insufficiency and dynamic airway obstruction
    b. Valvular and ischemic heart disease, restrictive cardiomyopathy
    c. Hepatomegaly and splenomegaly
    d. Careful positioning due to stiff joints, skeletal abnormalities, and peripheral nerve palsies
- Avoid succinylcholine if myelopathy/ spinal cord compromise
- Possible difficult iv access secondary to subcutaneous deposition of mucopolysaccharides
- Developmental delay – cooperation and consent issues

ANESTHETIC GOALS:

- Consider awake FOB intubation if concerned about difficult airway, or have a difficult airway approach keeping in mind minimizing manipulation of C-Spine.
- Thorough pre-operative work-up and optimization of possible co-existing cardiopulmonary conditions and organ function.

HISTORY AND PHYSICAL

- Variable degree of mental retardation frequently occurs
- Vision abnormalities secondary to corneal opacities and glaucoma
- Throrough history for symptoms of upper airway obstruction and neck instability, plus a thorough airway exam:
    - Macroglossia, floppy epiglottis, cervical spine instability secondary to hypoplasia of the odontoid process (Morquio syndrome), infiltrative deposits in laryngeal inlet and pretracheal tissues - Difficult visualization of larynx
    - Coarse facial features (gargoylism) and midface hypoplasia - Poor mask fit
    - Narrowed airway passages and redundant airway tissue – Difficult BMV and laryngoscopy
- History of copious and thick secretions, recurrent respiratory infections?
- Symptoms of dyspnea, exercise intolerance, chest pain, previous MI, murmurs on P/E
    - Restrictive cardiomyopathy associated with infiltration of the myocardium; results in diastolic dysfunction and diminished stroke output
    - Valvular and ischemic heart disease
    - Respiratory insufficiency from chest deformity
- Heart, liver, and spleen enlargement resulting from mucopolysaccharide accumulation
- Neurological exam for symptoms of myelopathy and spinal cord compromise (secondary to severe hypoplasia or absence of the odontoid process of C2 vertebra)
- Peripheral nerve palsies such as carpal tunnel syndrome
- Associated skeletal abnormalities such as lumbar lordosis, stiff joints, chest deformity, dwarfing, and limitation of joint motion

INVESTIGATIONS

- CBC, lytes, BUN, Cr, LFTs?
- ECG and Echo to evaluate ventricular function, valvular function, and the presence of infiltrative disease causing conduction abnormalities
- CXR for respiratory infection, PFT +/- ABG for obstructive or restrictive respiratory disease

OPTIMIZATION

- Endotracheal intubation may require fiberoptic assistance, and the anesthesiologist should have multiple plans for airway management
- May consider judicious pre-op sedation if uncooperative secondary to mental retardation (but beware over-sedating because of difficult airway and possible limited respiratory function due to restrictive disease)

ANESTHETIC OPTIONS

- Local / Regional / GA – depends on the procedure and underlying cardiopulmonary function

ANESTHETIC SETUP

- Difficult airway cart, set-up for FOB intubation
- Airway topicalization equipment
- Consider having heliox and steroids ready
- Standard emergency drugs

MANAGEMENT OF ANESTHESIA

Induction

- Will most likely require awake FOB intubation
- The use of a laryngeal mask airway as a rescue device may not be successful
- Avoid succinylcholine if spinal cord compromise on history and physical
- Careful head and limb positioning

Maintenance

- Whatever you want

Emergence

- Considerations for extubating a difficult airway

DISPOSITION & MONITORING

- Depends on the underlying cardiopulmonary function and the procedure

COMPLICATIONS

- Dynamic airway obstruction

PATHOPHYSIOLOGY

- The mucopolysaccharidoses are a group of disorders with enzyme defects that result in incomplete degradation of glycosaminoglycans
- The mucopolysaccharides are polysaccharides that yield mixtures of monosaccharides and derived products after hydrolysis
- Mucopolysaccharides are found in all cells
- The mucopolysaccharidoses are genetically determined diseases in which mucopolysaccharides are stored in tissues in abnormal quantities and excreted in large amounts in the urine. The disorders result from a deficiency of a specific lysosomal enzyme that is required to break down these compounds. As a result, mucopolysaccharides accumulate in tissues, producing specific clinical manifestations.
- There are seven basic forms of mucopolysaccharidoses and several subgroups
- Most of the mucopolysaccharidoses are inherited as autosomal recessive traits
- All the mucopolysaccharidoses are progressive
- Some cases have been successfully treated by bone marrow transplantation at a young age
- The Hunter and Hurler syndromes are the best-known variants of the mucopolysaccharidoses
    - Hunter syndrome is an X-linked recessive disease; often die at a young age secondary to respiratory and cardiac complications
- Mucopolysaccharidosis IV (Morquio syndrome) is associated with perhaps the most significant skeletal deformities

CONSIDERATIONS IN PREGNANCY

- Nothing in Chestnut on this disease

REFERENCES

- Cote Chpt 12, 22, 29, 32, 53
- Barash P. 617-618