Down's Syndrome – Trisomy 21

Down's syndrome is a genetic disease resulting in a broad range of phenotypes with multisystem implications.

ANESTHETIC CONSIDERATIONS:

1. Possible difficult airway; prone to airway obstruction
    a. Large tongue, large tonsils/adenoids, AA/AO instability, small mouth, sublottic stenosis
2. Atlantoaxial/Atlantooccipital Instability requiring C Spine precautions
3. Multisystem disease
    a. CNS: AA/AO instability, cord compression, developmental delay, hypotonia and possible aspiration risk
    b. CVS: AVSD, ASD, VSD, PDA, TOF with possible development of pHTN
    c. RESP: OSA, recurrent aspiration, subglottic stenosis
    d. GI: TEF, duodenal atresia
    e. Endo: hypothyroid, DM
    f. Heme: immune deficiency, increased risk of hematologic malignancy, anemia
4. Possible uncooperative patient
5. Obesity – OSA, difficult IV access
6. Age appropriate anatomic, pharmacologic and physiologic considerations.

ANESTHETIC GOALS:

- Preoperative optimization: EMLA, premedication (caution with OSA and respiratory depressants)
- Assess for systemic manifestations of Downs Syndrome
- Anticipate perioperative airway obstruction.
- Postoperative stridor.

PATHOPHYSIOLOGY

- Down syndrome (DS) is the most common chromosome abnormality among live-born infants, with a incidence of 1/700-1000 live births. It can be due to an extra chromosome 21, an unbalanced Robertsonian translocation (extra long arm of chromosome 21 on chromosome 14) or a mosaic of normal and extra chromosome 21s
- Almost all individuals with DS have cognitive impairment, although the range is wide
    - Most are mildly to moderately mentally retarded, with IQ in the 50 to 70 or 35 to 50 range, respectively, although some are severely impaired with IQ 20 to 35
- Approximately 40% of individuals with DS have congenital heart disease
- **Neuro / Behavioral:**
    - May have symptoms from C1-C2 subluxation (atlanto-axial instability—AAI, due to ligamentous laxity)
    - Microcephaly, dysplastic ears with frequent OM and conductive hearing loss, and upslanting eyes with strabismus
    - Wide spectrum of developmental delay, precocious Alzheimer's disease
- **Airway:**
    - Large protruding tongue, increased salivation
    - Large tonsils and adenoids, floppy soft palate and possible sleep apnea
    - Micrognathia with small, easily damaged teeth, or mandibular protrusion
    - Narrow nasopharynx/choanal atresia
    - Midface hypoplasia with high arched palate or palatal abnormalities
    - Flat occiput
    - 20-25% have relative subglottic stenosis
    - 20% have asymptomatic atlantoaxial instability, short neck
    - Intubation not usually difficult
- **Resp:**
    - OSA and central sleep apnea
    - Recurrent aspiration pneumonia
    - Chronic upper airway obstruction may lead to cor pulmonale and pulmonary hypertension
- **CVS:**
    - 40-50% with congenital heart disease
        - AVSD (also called endocardial cushion defect) with or without other lesions – 40-50%
        - Ventricular septal defect with or without other lesions – 27%
        - Isolated secundum atrial septal defect
        - Isolated persistent patent ductus arteriosus - 12%
        - Isolated tetralogy of Fallot - 8%
    - CAD in adult Down's
    - Frequent bradycardia under anesthesia
- **MSK:**
    - General hypotonia
    - Xerodermia, atopic dermatitis and obesity make IV access difficult
- **Heme:**
    - 1% incidence of leukemia (10-20X normal) AML or ALL
- **Endocrine:**
    - Hypothyroidism (increasing frequency with age)
    - Prone to hypothermia
    - Diabetes in adults
- **GI:**
    - GERD, Duodenal atresia, TEF, imperforate anus and Hirshsprung's disease

HISTORY & PHYSICAL EXAM

- History from parent/caregiver and old chart (cardiac disease, prior operations, pulmonary HTN, sleep apnea, lung function, recurrent aspiration pneumonia, adverse effects from prior anesthetics, airway)
- Complete functional enquiry (exercise tolerance, last URTI, neurological compromise or dysfunction suggesting AAI and cord compromise)
- Careful history to elicit neurological compromise: gait abnormalities, increased clumsiness, complaints of numbness, tingling in an extremity, weakness of an extremity, or a preference for the sitting rather than activity—may indicate AAI/spinal canal stenosis & cord compromise
- Careful airway exam: tongue hypertrophy increases with age—previously normal airway may be now difficult. Look for evidence of shunts / flow reversal / HF: listen for loud S2, S3 gallop murmurs; O2 sats and cyanosis, pulmonary edema. Assessment of spinal cord: hemiparesis or ataxia, hyperreflexia, clonus, neurogenic bladder, or sensory loss. Any of these signs or symptoms increase risk of dislocation, and mandate that a neurosurgical or orthopedic consult be made unless emergent surgery.

INVESTIGATIONS

- **Labs:** dependant on the surgery: CBC and differential, electrolytes, TSH, renal function
- **Imaging:** ECHO/ EKG/ CXR dictated by procedure and patient history
    - X-xray of neck controversial ? plan C-spine precautions for all

OPTIMIZATION

- Preoperative EMLA, sedation/antisialogogue (midaz may have unpredictable effect, IM ketamine)
- SBE prophylaxis (unrepaired cyanotic HD or within 6mo and not endothelialized) for dental procedures

ANESTHETIC OPTIONS

- Commonly used inhaled or intravenous techniques of general anesthesia are acceptable, but must be tailored for the presence of CHD
- Regional anesthesia not usually used due to behavioral issues and risk of hypoventilation with sedation

ANESTHETIC SETUP

- **Drugs:** standard emergency drugs; increased sensitivity to atropine with resultant mydriasis and profound tachycardia according to Coexisting, disproved according to Miller; consider milrinone/nitric oxide if severe PHTN
- **Equipment:** Ensure pre-oxygenation, prepare for difficult IV, apply monitors including 5-lead EKG
    - Need difficult A/W equipment and other adjuncts (i.e. intubating LMA)
        - Have a number of ETT with smaller sizes available due to the increased incidence of subglottic stenosis
    - Art line, TEE, central venous access dictated by patient (CHD) and procedure

MANAGEMENT OF ANESTHESIA

- **Induction:** balance potentially uncooperative patient with patient condition (ie avoid hypoxemia if sleep apnea/pulmonary HTN). Care re AAI. Vigilance re air in lines if CHD. Use ketamine for induction if minimal cardiac reserve. Bradycardia during induction is more common.
- **Maintenance:** either inhalational or intravenous, maintain normocarbia, avoid hypoxia, sympathetic stimulation if pulmonary hypertension.
- **Emergence:** Need full reversal if NDMR used as hypotonic. Judicious use of opiods if OSA, use of adjuvant analgesic medication. Early family presence to help calm child.

DISPOSITION & MONITORING

- Depends on surgery and patient history / co-morbid diseases

COMPLICATIONS

- 2% postintubation stridor
- Dental damage b/c teeth small with conical roots
- Infection due to ↓ immune competence
- Spinal cord compression from AAI
- High rate of postoperative morbidity and mortality following cardiac surgery d/t respiratory complications because of airway abnormalities (frequent postoperative pneumonia, atelectasis and pulmonary HTN)

REFERENCES

- Miller 7th edition, Barash 6th ed, Stoelting's Coexisting Diseases 5th edition