

Achondroplasia

Achondroplasia is the most common cause of disproportionate dwarfism. A decrease in the rate of endochondral ossification results in short tubular bones. This syndrome has important anesthetic implications including: possible difficult airway, cervical spine instability, foramen magnum stenosis - hydrocephalus, central sleep apnea, OSA, restrictive lung disease and pulmonary hypertension.

ANESTHETIC CONSIDERATIONS:

- Possible difficult airway
 - Anatomic changes: Large protruding forehead, short maxilla, large mandible, large tongue, cervical kyphosis and reduced neck ROM, proportionately smaller airways without anatomic abnormalities (pediatric ETT size should be weight based)
 - Difficult BMV and face mask fit
 - Large tongue, OSA and upper airway obstruction
 - Short laryngoscope, range of blades and oral/nasal airways should be readily available
- Atlantoaxial instability, atlantooccipital fusion and odontoid hypoplasia
 - Avoid neck hyperextension
- Foramen Magnum stenosis and brainstem compression
 - Avoid neck hyperextension (brainstem compression)
 - Central sleep apnea
- Hydrocephalus and increased ICP
- Pulmonary Hypertension and cor pulmonale
 - Kyphoscoliosis and associated RLD
 - Large tongue and redundant tissue: OSA
- Difficult IV access (excess skin and subcutaneous tissue)
- Anatomic abnormalities of the spine:
 - Difficult epidural: small epidural space
 - Difficult spinal: bony abnormalities
 - Dose adjustment of neuraxial LA

ANESTHETIC GOALS:

- Preoperative optimization and investigation
 - ICP
 - Pulmonary hypertension
- Anticipate possible difficult airway and possible difficult BMV
- Cautious C spine manipulation
- Avoid increases in pulmonary vascular resistance
- Avoid increases in ICP

HISTORY

- OSA
 - Snoring, apneas, daytime hypersomnolence, behavioural changes, irritability, AM headaches
- Central sleep apnea
 - May be a sign of brainstem compression from foramen magnum stenosis
- ICP
 - Headaches, visual changes, postural instability and imbalance
- Pulmonary Hypertension
 - Functional capacity

PHYSICAL

- **AIRWAY:** neck ROM, mouth opening, degree of scoliosis, MP (large tongue), sleep apnea?, facial abnormalities (ease of BMV?)
- **CNS:** signs and symptoms of increased ICP, signs and symptoms of foramen magnum stenosis and brainstem compression, atlantoaxial instability
- **CVS:** signs and symptoms of pulmonary hypertension and cor pulmonale
- **RESP:** kyphoscoliosis and associated restrictive lung disease
- **ABDO**
- **MSK:** redundant soft tissue and skin, IV access

INVESTIGATIONS

- Labs: routine, as indicated for surgical procedure
- ECG: pulmonary hypertension and cor pulmonale: RAD, p pulmonale, RBBB, R>S in V1/V2
- Echo: RVSP, pulmonary pressure estimation
- Sleep study

OPTIMIZATION

ANESTHETIC OPTIONS

- **None**
- **MAC with regional**
- **Neuraxial**
 - May be technically difficult
- **General**

ANESTHETIC SETUP

- **** Difficult airway cart**
- Standard machine setup
- Standard emergency drugs

MANAGEMENT OF ANESTHESIA

- **Induction**
 - Consider awake fiberoptic intubation due to increased risk of difficult airway and difficult BMV with C spine instability
- **Maintenance**
- **Emergence**

DISPOSITION & MONITORING

OBSTETRICS

- C section necessary because small contracted pelvis combined with near normal birth weight infant = CPD
- Regional anesthesia may be technically difficult
 - Dose adjustment of neuraxial LA
 - Epidural may be preferable to spinal to allow for appropriate dose adjustment

PATHOPHYSIOLOGY

- The most common cause of disproportionate dwarfism
- Incidence: 1.5:10000 births
- Autosomal dominant transmission (~80% of cases represent spontaneous mutations)
- Basic defect: decrease in rate of endochondral ossification; coupled with normal periosteal bone formation → short tubular bones
- Mean height: males – 132 cm; females – 122 cm
- Signs and symptoms:
 - Normal mental and skeletal muscle development
 - Kyphoscoliosis and genu varum (bow legs) are common
 - Premature fusion of bones at skull base can result in shortened skull base and foramen magnum stenosis
 - Functional fusion of atlanto-occipital joint with odontoid hypoplasia, atlantoaxial instability, bulging discs and severe cervical kyphosis
 - Hydrocephalus
 - Cervical spinal cord compression and damage
 - Difficult airway due to C spine kyphosis (and airway/facial abnormalities)
 - Central sleep apnea may result from brainstem compression due to foramen magnum stenosis
 - Obstructive sleep apnea may result from large tongue and facial abnormalities
 - Pulmonary HTN may develop secondary to RLD and severe kyphoscoliosis or ongoing central/obstructive sleep apnea
- Normal life expectancy if survives 1st year of life
- Surgical presentations:
 - **SUBOCCIPITAL CRANIECTOMY FOR FORAMEN MAGNUM STENOSIS**
 - **LAMINECTOMY (SPINAL STENOSIS OR NERVE ROOT COMPRESSION)**
 - **VP SHUNT**
 - **C SECTION**

REFERENCES

- Anesthesia and Coexisting Disease 5th Edition