Anesthetic Considerations for Pediatric Patients with Achondroplasia

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Learning Objectives

• Describe the pathophysiology of Achondroplasia
• Recognize the clinical features and comorbidities associated with achondroplasia
• Discuss preoperative evaluation of pediatric patients with achondroplasia
• Develop an appropriate anesthetic plan to care for pediatric patients with achondroplasia
What is Achondroplasia?

• “A-khondros-plasisia”: no Cartilage growth
• Ancient Egypt around 2700 BC
• 1/20,000 live births
• Most common cause of short stature or “dwarfism”

Wellcome Collection gallery (2018-04-03): https://wellcomecollection.org/works/cgpb7a6c CC-BY-4.0
Inheritance

20%

Autosomal dominant

Affected father (mutated gene)

Unaffected Mother

Affected son (mutated gene)
Unaffected daughter
Unaffected son
Affected daughter (mutated gene)

Probabilities: 1:2

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Point Mutation: (80%)

• Associated with increased paternal age
• New point mutation in the Fibroblast Growth Factor Receptor 3 gene (FGFR3)
Pathophysiology of Achondroplasia

Over-activation of the FGFR3 receptor expressed in chondrocytes and mature osteoblasts

- Inhibition of growth plate size and cartilage proliferation and premature ossification of epiphyseal cartilage
- Impaired bone growth during childhood

https://commons.wikimedia.org/wiki/File:608_Endochondral_Ossification.jpg
Achondroplasia: Common Surgeries and Procedures that Require Anesthesia

• Magnetic Resonance Imaging of the spine and brain
• Decompressive sub-occipital craniectomy
• Cerebrospinal fluid shunting procedures
• Adenotonsillectomy & myringotomy tube placement
• Various orthopedic procedures including limb lengthening, laminectomies, and kyphoscoliosis correction.
Clinical Features: Craniofacial

- Large head
- Frontal bossing,
- Midface hypoplasia
- Large tongue
Clinical Features: CNS

- Normal intelligence
- Delayed motor development
- Cervical medullary compression
- Cervical myelopathy
- Hydrocephalus
- Psychosocial problems
Ten percent of affected individuals suffer from true cervical medullary compression requiring surgical decompression and 2-5% experience sudden infant death in the absence of aggressive evaluation.
Clinical Features: ENT

- Recurrent otitis media
- Risk of hearing loss
- Language and speech articulation problems
- Learning disabilities.
- Both central apnea and obstructive sleep apnea

https://commons.wikimedia.org/wiki/File:Blausen_0328_Ear
Sleep Disordered Breathing (SDB)

Central Sleep Apnea
- Medullocervical compression

Obstructive Sleep Apnea
- Compression of the lower motor neurons
- Altered craniofacial features
- Small upper airway
- Physiologic adenoidal hypertrophy
- Hypotonia of nasopharyngeal airway muscles

*Infants with achondroplasia have attenuated arousal response which could play a role in SIDS*
Clinical Features: Cardiopulmonary

• Restrictive Lung Disease

• Pulmonary hypertension

• Cor-pulmonale
Clinical Features: Gastrointestinal

*Children with Neuro-Respiratory Complications:*

- Severe Gastroesophageal Reflux (GERD)
- Hyper salivation
- Chronic aspirations
Clinical Features: Musculoskeletal

*Thoracolumbar kyphoscoliosis*
- Worse during infancy
- Improves with age

*Lumbar lordosis*
- Worsens with Age
Clinical Features: Musculoskeletal

- Rhizomelic Shortening
- Brachydactyly
- Genu varum
Preoperative Evaluation

**Preoperative history and exam should focus on:**

- **Airway:** potential difficult ventilation and intubation
- **Cardiopulmonary Status:**
  - Restrictive lung diseases,
  - pulmonary hypertension
  - Cor-pulmonale
- **Neurological Status**
  - Baseline neurological exam
  - Consider baseline neuro-imaging
Preoperative Medication

• Anxiolytics : Emotional and Physical Stress
• Anti-sialagogues : Helps with Fiberoptic intubation (FOI).
• Histamine H₂ antagonists : GERD patients
Perioperative Management

Monitors

• ASA standard monitoring.
• Challenging BP cuff size
• Invasive monitoring:
  - Surgical procedure
  - Patient’s baseline cardiopulmonary function.
Perioperative Management

• **Difficult Peripheral Access:**
  - Short proximal extremities
  - Redundant soft tissue
  - Increased subcutaneous fat
  - Contracted joints

• **Difficult Central Access:**
  - Short neck and large head: difficult internal jugular access
  - Narrow deformed chest: difficult subclavian access
  - Contracted Hips: difficult femoral

*Consider vein viewer, ultrasound and intraosseous*
Perioperative Management

**Positioning**
- Knee joints are often very lax
- Hip & elbow Joints often have limited mobility and contractures

**Temperature**
- Head is relatively large and patients can lose heat through their head
- Warm the operating room and cover the head
Pediatric patients with achondroplasia may have difficult airways to manage

• Review airway management on previous anesthetic records
• Maintain spontaneous ventilation if possible
• Ventilatory difficulty with chronic lung disease patients
• Mask ventilation may be challenging
• Supraogglottic airways may not fit well and a variety of sizes and types should be available
Airway Management

Caution should be exercised while manipulating the neck during intubation

- Foramen magnum stenosis
- Odontoid hypoplasia
- Cervical kyphosis
- Cervical neck fusion
- Large tongue
- Narrow nasopharynx

Recommend In line stabilization and Video-laryngoscope or Fiberoptic bronchoscope for intubation
Intraoperative Management

• Intravenous and inhalational anesthetic agents
• Medication dosages based on weight.
• Multimodal analgesia with sleep apnea.

• For patients with cardiopulmonary compromise:
  - Maintain blood pressure
  - Maintain adequate tidal volume
  - Avoid hypoxia and hypercarbia
Regional Anesthesia

- Epidural catheter
- Spinal single injection
- Single caudal injection
- Caudal catheter
- Peripheral nerve blocks
Conclusions

• Autosomal dominant disorder caused by mutations in the fibroblast growth factor receptor 3 (FGFR3) gene
• Achondroplasia presents with unique craniofacial, short stature with disproportionate trunk to limb ratio due to rhizomelic shortening of the limbs.
• Life threatening in 5 - 10% of affected individuals.
• Preoperative anesthetic evaluation should focus on assessment of airway, neurological and cardiopulmonary function
• With appropriate knowledge and proper care, anesthesia can be delivered safely to pediatric patients with achondroplasia
References


• Di Nardo SK. Anesthetic consideration for the achondroplastic dwarf. *AANA J* 1988 Feb;56(1):42-