Skeletal Dysplasia

Mary Theroux, MD
Pediatric Anesthesiologist, Director of Research
Alfred I. DuPont Hospital for Children
Professor of Anesthesiology and Pediatrics
Thomas Jefferson University
Philadelphia, PA

Disclosures

- Financial - None
- Member of Skeletal Dysplasia Management Consortium
  (Sole Anesthesiologist among ENT/ Pulmonology/Orthopedics– Lonely Job)

Definition of Conference

“The confusion of one man multiplied by the members present”

Anonymous

Goals

- Briefly discuss SD of concern
- Not all can be discussed – simply too many
- Discuss common perioperative concerns
- Where the pitfalls likely occur
- New Information available on MPS


- ~436 disorders involving 364 genes (Bonafe L et al)
- Disproportionate Short stature – defined by height that is 3 or more standard deviations below the mean height for age
- If short stature is proportional: may be due to endocrine or metabolic disorders or chromosomal or non skeletal dysplasia (Chen et al: 2015)

Which SD to watch out for?

SD likely to require cervical spine fused early in life

- Progressively become difficult airway
- Metatropic and Kniest Dysplasia (MD)
- Spondyloepiphyseal Dysplasia (SED)
- Diastrophic Dysplasia (DD) 1: 500,000 (Finnish)
More such patients……………

- Campomelic Dysplasia
- Mucopolysaccharidosis especially IVA and VI
  - (Morquio – Brasilford Syndrome; Maroteaux – Lamy syndrome)
- Osteogenesis Imperfecta (will not cover in as great detail - greater awareness; more common)

Osteogenesis Imperfecta

- May have difficult airway
- Fractures here, there and everywhere…

Osteogenesis Imperfecta :MHS?

- More recent publications suggest that the hyper metabolic state and hyperthermia observed in OI during the peroperative period are not MH related
- Because they are self-limiting, muscle rigidity is not seen, and normocarbia is maintained. (Baum et al, Cho et al)

Other Practical Difficulties

- Obvious One - fear of fractures
  - Have surgical team help transfer from bed to bed (share the blame…)
  - Blood Pressure measurements
    - Less often
    - Use pre-cordial stethoscope and document
    - Use neonatal cuff
    - Arterial line – not unreasonable escalation of care
    - IM roding of long bones – bleeds………..

Achondroplasia

- Incidence .36 -.6 per 10,000 live births
- Often NOT difficult airway until later in life.
- Incidence of difficult airway unknown but adult with Achondroplasia is much more likely difficult
- Foramen magnum stenosis common
  - NO instability of cervical spine
- Thanatophoric Dysplasia 0.21 – 0.3 per 10,000
Achondroplasia

- Foramen Magnum Stenosis
  - rarely seen in other skeletal dysplasias
  - important to recognize, evaluate and treat early in life

- Infants may need MRI of brain and spine
- Foramen magnum stenosis may lead to
  - Apnea
  - Poor feeding
  - Sudden death
- Watchful waiting is most desired management unless severe
- Most infants improve with time and growth

Teenage – Achondroplastic

- Lumbar stenosis common
- In general uncomplicated anesthetic
- Temperature likely to rise especially if temperature conservation applied
  - Other skeletal dysplasia do this as well
  - Parents give history of sweating and intolerance to heat – kick their blankets off
- Complications possible

Moving on: High Risk groups

- Relatively new information
- Will cover what has been 'simmering' in the LPA (Little People’s Association)
- Two main issues
  - Airway: One would think 'No real surprise there', No longer true
  - Spinal cord infarction: More in MPS but others at risk as well?

News of loss of airway occurring all over the country

- 20+ year old spondylo metaphyseal dysplasia
- Breast reduction – cannot intubate – difficulty ventilating - tracheostomy – died in ICU within a week
- 22 year old Morquio patient – c/o respiratory difficulty – Her primary care physician thought she was developing allergies since it was early spring – gave antihistamines? Found dead in bed
- Is this just expected mortality in this population?

Mucopolysaccharidosis

- Been in the medical news and social media

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Mucopolysaccharidosis

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M.S. Muhlebach et al. / Paediatric Respiratory Reviews 12 (2011) 133–138
“There are no minor anesthetics for MPS patients even when surgery is minor”

Clinical course of a 16 year old MPS IVA patient, height: 90 cm; body weight: 16 kg at 16 years old)

Progressive deformity of the trachea on radiographs

- With age, the trachea takes on a tilted hourglass configuration in the frontal projection with narrowing and deviation to the right at the thoracic inlet.
- Tracheal narrowing in the AP direction is often difficult to see on lateral radiographs due to overlapping shoulders.

Findings from Ct / Angio

Surgical reconstruction for severe tracheal obstruction in Morquio A syndrome: Pizzari, Davies, Theroux, Spurrier, Averill, Tomatsu, Paul Harmatz: IN PRESS

CTA (chest) - 16yo boy with Morquio A

- He was experiencing increasing fatigue, respiratory distress not relieved with CPAP, with moderately severe obstructive lung disease and sleep apnea
- Receiving enzyme replacement therapy
- After clinical and imaging work-up, underwent tracheal reconstruction with end-to-end anastomosis and translocation of the BCA to the proximal ascending aorta
- Improved pulmonary symptoms and PFTs after surgery

Sagittal and coronal reconstructions from CTA of the chest show marked narrowing of the trachea (arrows) with deviation rightward at the thoracic inlet. The BCA (arrows) has a leftward origin from the aortic arch, and then takes a tortuous course, passing anterior to the trachea. The BCA causes marked anterior compression of the trachea.

Chondrocytes in trachea (light microscopy). Arrows show chondrocytes filled with vacuoles. Left panel: hyaline chondrocytes and fibrous chondrocytes.
MR of the cervical spine – Tracheal evaluation

- Addition of coronal plane is useful to show multiplanar nature of tracheal narrowing and deviation at the thoracic inlet

Same boy, age 8. Coronal T2-weighted images show focal narrowing of the trachea at the thoracic inlet (arrow) with deviation to the right. The trachea is normal caliber above and below this level (arrow) with crossing tortuous BCA (arrow). Clinically, he was experiencing increased shortness of breath with activity as well as heavy, noisy breathing.

[Video link]

CTA of the chest – 24yo woman

Airway fly through correlates with the 2D representation and bronchoscopy. Bronchoscopy images were limited with patient under limited sedation. Note that by convention, patient’s right is displayed on image’s right for airway fly through; to match conditions of bronchoscopy. Due to difficulty ventilating, though, bronchoscopy camera orientation was altered from the norm.

[Video links]

MR of the cervical spine – Tracheal evaluation

- Trend towards worsening narrowing with neck flexion
- Anterior impression on the trachea by the crossing tortuous brachiocephalic artery (BCA) was seen in 15/19 patients with tracheal narrowing
- All patients older than 15yo had at least 50% narrowing
- Trend towards worsening narrowing with neck flexion

34 MR exams of the cervical spine in 28 children and young adults with Morquio A showed age-dependent tracheal narrowing in neutral neck positioning.

Scale: 0 = no narrowing, 1 = greater than 25% narrowing, 2 = 50-75% narrowing, 3 = greater than 75% narrowing.

4th MR exam age in 28 children and young adults with Morquio A showed significantly worsening tracheal stenosis with age (p<0.001, one-way ANOVA).

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4th MR exam age in 28 children and young adults with Morquio A showed significantly worsening tracheal stenosis with age (p<0.001, one-way ANOVA). Scale: 0 = no narrowing, 1 = greater than 25% narrowing, 2 = 50-75% narrowing, 3 = greater than 75% narrowing.

Tracheal narrowing used for reference.

Patients 1 through 4 subjected to tracheal resection and anastomosis in addition to innominate artery relocation (except pt#2). All had severe narrowing of trachea at thoracic inlet (arrow) with compression of trachea by innominate artery (except pt#2). Pt#3 had severe stenosis of trachea close to the carina (arrow). Pt#4 through 8 awaiting evaluation.

[Video links]

Four patients have been subjected to cardiothoracic surgery

<table>
<thead>
<tr>
<th>Patient</th>
<th>Preoperative presentation</th>
<th>Hospital course</th>
<th>Postoperative complications</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Moderate airway obstruction, GORE expanded polytetrafluoroethylene (ePTFE) tracheal stent</td>
<td>Moderate airway obstruction, GORE ePTFE tracheal stent</td>
<td>None</td>
</tr>
<tr>
<td>2</td>
<td>Severe airway obstruction, tracheal stent</td>
<td>Severe airway obstruction, tracheal stent</td>
<td>None</td>
</tr>
<tr>
<td>3</td>
<td>Severe airway obstruction, tracheal stent</td>
<td>Severe airway obstruction, tracheal stent</td>
<td>None</td>
</tr>
<tr>
<td>4</td>
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</tbody>
</table>

[Video links]
What is the other HOT topic?

- Dense irreversible spinal cord infarction following non–spine surgery
  - 29 year old who had sustained T4 spinal cord infarction during a C1 laminectomy
  - The point is that it did not reach the average Anesthesiologist or Orthopedic surgeon. In fact still has not been quoted

They discuss ventilation difficulties

Tracheostomy tube obstruction – recurrent episodes. Below are view through FOB. Not surprising given what we know now

More Recently


- More cases around the country
Points to consider

- Doubt that epidural anesthesia per se is directly injurious to the spinal cord of patients with Morquio A
- Two of the authors (Theroux, Krane) have employed epidurals to good effect in these patients.
- Nonetheless, because of the apparent vulnerability of Morquio A patients to spinal cord injury, we now view epidural anesthesia as relatively contraindicated for a number of reasons.

Recommendations

- We considered the possibility that an epidural infusion might increase neuraxial pressures and decrease local perfusion pressure. However, the intervertebral foramina were reported to have been patent.
- In patients with Morquio A syndrome, it seems prudent to avoid epidural anesthesia, to provide careful support of BP, and to avoid flexion. In the event that cord imaging is not available, e.g., emergency procedures, it would be prudent to assume the presence of spinal stenosis (as well as atlantoaxial instability).
- In addition, intraoperative electronic monitoring of the spinal cord should be considered, especially if the status of the spinal cord cannot be confirmed prior to surgery.

So now what about the spinal cord infarction?

- There were six epidurals successful of eight attempted.
- 4/6 were caudally placed. Caudal age 8.6 years (range, 6–10 years).
- Lumbar approach was 11.2 years (range, 8–14 years).
- Caudal approach was preferred because of known irregularities of the vertebral bodies (anterior beaking and pelyndromelia) and frequently present thoracolumbar kyphosis. The successful catheter placement resulted in satisfactory pain control.

All MPS patients at risk?

- We think so. Why? Well.............
- Some things have no answer: Why isn’t the number 11 pronounced onety-one?
- MPS I, MPS IVA, MPS VI: We have cases that we know of.
- One – MPS I (Hurler’s)
- Four – MPS IVA (Morquio)
- One – MPS VI (Maroteaux – Lamy)

Metatropic Dysplasia

- Metatropic Dysplasia—Little People with Challenging Airways: How Can We Reduce the Risk? Theroux et al: IN REVIEW, Pediatric Anesthesia

Skeletal Dysplasia = Difficult Airway

- Have to discuss airway and intubation, or else I am not doing my job.

Skeletal Dysplasia
Frequency of use of difficult airway tools. Glidescope was the most preferred tool as it allowed displacing oropharyngeal structures effectively. When Miller or Macintosh blades were used with success the intubations were noted to be more difficult with multiple attempts.

Entire cervical spine is fused (spontaneously) even though surgically the spine was only fused from Occiput to C2. Crowding of structures in the neck including the prominently seen sternal heads of the clavicle. Cricoid is not palpable in this patient as it is located below the sternal notch.

What went wrong the last minute?

Retraction of the tongue using a piece of gauze followed by placement of Glidescope blade.

Problems with reinforced ETT

Tongue Retractor

- Can be life saving
- Often forgotten during emergencies
- Difficult airway cart
Results: Difficult Airway Letter

**Ventilation with face mask**

Difficult: Need two person assisted ventilation. It can be critical that the ventilation is performed with a second mask in place. This is also necessary if tongue is retracted and not in the oral cavity. Great discomfort will have the tip of the tongue actually protruding outside the oral cavity.

Intubation tools that failed:
- Guedel or flexable (which has been used before).
- May have been time of tongue was retracted due to inability to visualize a normal cartilage may be abnormal in that area. In deep intubation cases, there is a risk with these intubation devices, which may have been due to anesthetic associated requiring multiple attempts.
- Intubated using: An esophageal scope with full tongue retraction by an assistant. May also have been intubated with esophagus lying due to a recent severe bout of gastroesophageal reflux.

**Intubation using: F 7 esophageal scope 2.8 with full tongue retraction by an assistant.**

**Success of the day.**

Intubated using: F 7 esophageal scope 2.8 with full tongue retraction by an assistant. May also have been intubated with esophagus lying down due to a recent bout of gastroesophageal reflux.

Level of consciousness after intubation: Awake with brief period of apnea. Note that when the need for ventilation conditions and may have been intubated with in the area. Observed by assuring anesthesiologist present who needs to provide ongoing anesthetic, care. He does have advice on a number of occasions, and that intubation with esophagus lying down may have given more then, since, always the slightest thing can give the impression of complications.

Results

- **No neuraxial anesthetic**
- **Multimodal pain management including use of PCA**
- **Agents used:** Morphine, hydromorphone, clonidine, nalbuphine in selected cases, dexmedetomidine, clonidine patch, acetaminophen, ketorolac and gabapentin and diazepam

Metatropic Dysplasia - Results

- **Lack of familiarity with a particular patient’s airway even when managed by experienced anesthesiologists may result in a bad outcome.**
- **Patient whose airway had been secured either via use of GVL or flexible fiberoptic bronchoscope (oral route) when at a different hospital resulted in prolonged and traumatic intubation, the need for mechanical ventilation, ENT evaluation for damages incurred, extubation after 48 hours.**

**Results**

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**Guidelines for Extubation**

- **Attention to small details**
- **Be patient to allow complete emergence**
- **Vigilance for unanticipated respiratory difficulty**
- **Normothermia – If > 36 to 38**
- **Maintain child’s head and neck position similar to baseline**

**This note was on page # 102 of the chart sent by request**

Why not give an airway/anesthesia management letter? Most specialties do ……

**Cases reported to MHAUS meeting**

- **TITLE:** HYPERThERMIA IN CHILDREN WITH DWARFISH. Reported to MHAUS
- **AUTHORS:** MARY C. THEROUX, M.D and ROBERT G. KETTRICK, M.D.
  - A. I. duPONT CHILDREN'S HOSPITAL. WILMINGTON, DELAWARE.
- **We present two cases of hyperthermia in children with dwarfism.**
  - **Case 1**: Two year old white female. Dx of SED form of SD posterior cervical fusion for cervical spine instability.
  - **Anesthetic:** Isoforane, Sufentanyl and Vecuronium.
  - Axillary temperature at the start of the case - 96.8F (36.0C)
  - Progressively climbed to 100.2F (37.9C)
  - Approximately four hours later temperature rose to 105.5F (40.8C) rectally.
  - Patient was treated with tylenol and temperature progressively decreased to normal and stabilized. CPK was not elevated, electrolytes were normal and there was no metabolic acidosis present.

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**Skeletal Dysplasia**
Case # 2: Hyperpyrexia – Benign?

- Twenty month-old white female, Achondroplastic form of dwarfism for exploration of posterior fossa
- Esophageal temperature slowly increased intraoperatively over the next two hours to 104.6°F (40.3°C) with no accompanying tachycardia
- Lab investigations revealed no metabolic acidosis (base deficit 2.6), potassium 3.9, calcium 4.5 and CPK 134.
- The patient was given one dose of dantrolene 2mg/kg.
- Surgery was cancelled and patient was transferred to the intensive care unit. On arrival to the ICU, the patient's temperature was 99.6°F (37.5°C).
- Patient was extubated later the same day.

**Discussion:**
- Are some form of SD patients known to have hyperthermia in association with general anesthesia?
- We now know the answer: YES

People Change

- Difficult Intubation
- Surprised me last time
- Intubated using Glide scope without much difficulty ONLY A YEAR PRIOR

Take Home Points

- Short stunted disproportionate SD patients are difficult
- Two most important aspects
  - Airway
  - Spinal Cord
- Please provide communication regarding airway – Cannot emphasize enough

End of Slides