Introduction

- Menkes kinky hair syndrome: X-linked recessive disorder occurring in 1 in 100,000-250,000 newborns.
- A mutation resulting in abnormal copper transport leads to dysfunction of several organ systems with symptoms including:
  - Hypotonia
  - Poor pharyngeal muscle control
  - seizures
  - Developmental delay
  - Failure to thrive
  - Kinky hair, depigmentation
- Death typically occurs by 3 years of age.
- Association of congenital subglottic stenosis (SGS) with Menkes is previously undescribed in the English literature. A Japanese group in 2009 described a patient with Menkes and SGS.

Case History

- 12 month-old male
- Past medical history: Menkes disease diagnosed at 5 months of age, gastroesophageal reflux, recurrent aspiration and failure to thrive requiring nasogastric feeds, hypotonia.
- Past surgical history: none

Case Details

- Taken to the operating room for laparoscopic gastrostomy (G) tube placement
- Menkes disease diagnosed at 5 months of age, gastroesophageal reflux, recurrent aspiration risk due to recurrent aspiration
- Direct laryngoscopy resulted in a grade 1 view, however an appropriately sized ETT was unable to be advanced
- 2.5 uncuffed ETT was able to be placed with some resistance, no air leak at 30 cm H2O.
- ENT service performed direct laryngoscopy and bronchoscopy immediately following G tube insertion.
- Diagnosis of type III laryngeal cleft and grade 1 subglottic stenosis was made (Fig 1).
- Extubated and taken to PACU in stable condition.
- Required racemic epi overnight for stridor, discharged on POD #2

Two months later

- Returned for laparoscopic Nissen fundoplication.

- Again, a 2.5 uncuffed ETT had to be used.
- Extubated and taken to PACU. Received racemic ephedrine and supplemental oxygen. Also caffeine Smgp/kg for periods of apnea causing bradycardia to the 50s.
- In the PICU he received Heliox and HFNC overnight due to stridor and increased work of breathing.
- He required ICU monitoring for four days. Discharged home on POD #6

Discussion

Subglottic Stenosis [5-7]

- Classified as congenital or acquired. Only 5% of all cases are congenital.
- Congenital subglottic stenosis is defined as a subglottic diameter < 4 mm in a full-term neonate and < 3.5 mm in the premature neonate.
- Diagnosis depends on exclusion of acquired stenosis.
- Grading based on the Myer-Cotton grading system (Fig 3).

Anesthetic Considerations for Menkes [1-3,5]

Neurologic:
- Seizures can be difficult to control. Patients may present on several anti-epileptic medications.
- CNS degeneration. Consider avoiding depolarizing neuromuscular blockers
- Persistent hypothermia
- Hypotonia

Respiratory:
- Recurrent aspiration pneumonia/pneumonitis
- Poor control of pharyngeal muscles
- Small chin, prominent upper incisors and large cheeks

Gastrointestinal:
- Gastroesophageal reflux due to neurodegeneration

Cardiovascular:
- Theoretical increased risk of cardiac conduction disturbances, mitral valve regurgitation, vascular aneurysms and difficult IV access

Hematologic:
- Theoretical increased risk of bleeding

Conclusions

- Airway abnormalities in Menkes syndrome may include unrecognized subglottic airway stenosis and compromise. Providers should prepare for advanced airway management as well as appropriate postoperative monitoring prior to anesthetizing children with this disease.
- Consider planning for prolonged postoperative monitoring in an ICU setting.
- Ultimately this patient required significant perioperative interventions related to the management of Menkes syndrome sequelae and previously unrecognized subglottic stenosis. Increasingly invasive interventions, from steroid administration, racemic epinephrine, and caffeine, through Heliox and HFNC/CPAP, and up to postoperative intubation and tracheostomy, may be required.

References


Fig 1. Picture obtained by ENT at time of airway evaluation. Shows SGS.

Fig 2. Patient’s EKG revealing wandering pacemaker and supraventricular tachycardia.

Fig 4. Percent Obstruction of Laryngotracheal Stenosis Estimated by Endotracheal Tube Size [5]