What’s on the Horizon for Sleep and Down Syndrome

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What are some emerging treatment options?

• More access with Telemedicine
• Custom Fitting PAP masks
• High Flow Nasal Cannula
• Drug Induced Sleep Endoscopy directed surgeries
• Hypoglossal Nerve Stimulator
• Pharmacologic Treatments
Trisomy 21

• Most common chromosomal disorder in children
• Up to 60% of children with Down Syndrome have OSA – and it tends to be severe
• 50-80% have residual disease after adenotonsillectomy
Telemedicine in Pediatric Sleep

Shalini Paruthi, MD\textsuperscript{a,b,}\textsuperscript{*}


- Doctors should try synchronous or asynchronous modalities of telemedicine (phone, video, websites, audio recordings, social media, etc..)

- Use televisits when possible

- In person visits still important in evaluation of obstructive sleep apnea or CPAP mask fitting
Severe OSA

adenotonsillectomy

Moderate/Severe OSA

Drug Induced Sleep Endoscopy (additional targeted surgery)

PAP tolerating

Not tolerating

Consider Alternative Therapies
• Nasal pillows
• Nasal cradle
• Nasal mask
• Full face mask
• Total Face Mask

Figure 1. Nasal Pillows and Nasal Cradle Mask Examples (a) Nasal pillows PAP mask, sealing the nares. (b) Nasal cradle mask, sealing under the nares.

Figure 2. Nasal and Full Face Mask Examples (a) Nasal PAP mask, over the nose. (b) Full Face PAP mask covers nose and mouth.
Feasibility of three-dimensional facial imaging and printing for producing customized nasal masks for continuous positive airway pressure

Using 3D face scans and engineering technology, customized masks are more feasible.

Consider exploring how to obtain a customized mask if needed.
High Flow Nasal Cannula?

High flow nasal cannula for children with severe obstructive sleep apnea not compliant with continuous positive airway pressure

Alessandro Amaddeo, Sonia Khirani, Annick Frasin, Thao Tang, Lucie Giffon, Brigitte Feurolia
ERJ Open Research 2019 5; P132; DOI: 10.1183/23120541.sleepandbreathing-2019.P132

- Study: 8 patients (mean age 9yo, mean AHI 33±22)
- Results: after 1 month, 62% slept with HFNC 4+hrs, mean AHI 2±2
- But not tolerated in 3 older DS patients
- Conclusion: HFNC may be used as a rescue therapy for children not compliant with CPAP. Further studies are needed
High flow nasal cannula treatment for obstructive sleep apnea in infants and young children

- Retrospective review of treatment with heated humidified high flow nasal cannula (HFNC) as treatment for OSA in young children intolerant of PAP, or at high risk for midface hypoplasia
- 22 children with OSA (AHI 4.8-89.2 events/hr),
- 19 patients received HFNC at home
  - by 12 months, 5 discontinued due to intolerance, 3 had resolved OSA
  - Common complications included: skin irritation, dry mucous membranes, increased central apneas
- Can serve as a bridge to surgery or spontaneous resolution of OSA

High flow nasal cannula treatment for obstructive sleep apnea in infants and young children

- HFNC appears to be an effective treatment for OSA with increased tolerance and less risk for mid face hypoplasia.

- Prospective needed:
  - Explore ventilation support
  - Compare to CPAP
  - Understand/address adherence issues

Drug-induced sleep endoscopy directed surgery improves polysomnography measures in overweight and obese children with obstructive sleep apnea

- Retrospective analysis of PSG parameters following DISE-directed surgery in obese and overweight pediatric patients.
- 40 children (mean BMI 94% percentile, mean age 8).
  17 children had undergone previous T&A,
  Included children with comorbidities (Trisomy 21, developmental delay, etc…)
- Interventions performed included: T&A, supraglottoplasty, lingual tonsillectomy or a combination. 2 patients (prior T&A) did not have additional intervention

- DISE-directed interventions resulted in statistically and clinically significant improvements in PSG parameters. Improvement in oAHI, oxygen nadir, ODI noted.
• JAMA case series of 6 adolescents (12-18yo) with DS and severe OSA (AHI >10), after T&A.
• Device: Inspire
• Measures: AHI, QOL measures
• Conclusion: good tolerance, 56-85% reduction in AHI, clinical improvement, improved QOL
Update on Hypoglossal Nerve Stimulation in Children With Down Syndrome and Obstructive Sleep Apnea

- Hypoglossal Nerve Stimulator (HGN) stimulation
- Case series of 20 children with Down Syndrome, ages 10-21 yo
- All participants had severe OSA (AHI >10, <50/hr) who failed CPAP therapy. All s/p DISE to confirm candidacy
- Patients underwent PSG HGN titration with reduction of AHI of 85% (though AHI was not fully normalized)
- Median nightly usage 9.21 hr/night. Caregivers reported improvement in QoL (OSA-18)
- 2 patients had interval adverse events requiring revision surgery that resolved problem
- 2 other patients underwent device interrogation to optimize tongue protrusion during DISE

- HGN still appears to be a reasonable option for patients with severe OSA, at risk for severe sequelae from untreated OSA, who are intolerance of PAP therapy
- Further study continues
Novel Medications for OSA

Combination medication Atomoxetine (a norepinephrine reuptake inhibitor) and Oxybutynin (an antimuscarinic) Taranto-Montemurro, et al 2020

- Increase genioglossus muscle responsiveness (upper airway dilator muscle)
- Reduced OSA severity by 63% in adults

New Studies suggest Long term use (>1 year) is effective Chen, et al 2021

Clinical Trial of Medication ClinicalTrials.gov NCT04115878

- Children with Down Syndrome, ages 6-17 yo
- Comparing high dose and low dose
- Primary outcome: oAHI
- Secondary outcomes: OSA QoL measures, arousal index, caregiver impression, sleep architecture


Be willing to think outside the box with your Trisomy 21 patients...