Management of OSA in children with Down syndrome: the otolaryngologist’s perspective

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Objectives:

- Review the clinical features and common otolaryngologic manifestations of Down syndrome
- Discuss the mechanisms and predisposing factors for sleep-disordered breathing in children with Down syndrome
- Discuss the initial approach and management options for OSA in children with Down syndrome – as well as typical outcomes
- Identify treatment options and expectations for persistent OSA after T&A in children with Down syndrome
- Discuss emerging technologies and techniques for management of complex, persistent OSA in children with Down syndrome
Down syndrome manifestations

- Characterized by a variety of dysmorphic features, congenital malformations, and other medical conditions
  - Not all manifestations are present in each affected person
  - Variable impact of these manifestations (mild → profound)
- Manifestations impact nearly all body systems, including multiple and significant issues involving the head & neck
ENT manifestations of Down syndrome

- Ears/hearing
  - Narrow ear canals
  - Chronic middle ear infections/fluid
  - Inner ear abnormalities
- Chronic rhinosinusitis
  - Smaller sinuses and drainage pathways
  - Small nasal passageway
  - Immunologic deficiencies
ENT manifestations of Down syndrome

- **Sleep apnea**
  - Smaller midface/jaw
  - Small nasal passageway
  - Macroglossia (larger tongue), lingual tonsil hypertrophy, glossoptosis (backward collapse of tongue)
  - Small upper airway and medially positioned tonsils
  - Decreased tone
  - Obesity

- **Laryngotracheal anomalies**
  - Floppy supraglottis/laryngomalacia
  - Complete tracheal rings
  - Narrowing below vocal cords
  - Tracheal bronchus

- **Dysphagia**
  - Hypotonia
  - Motor coordination
  - Reflux
OSA prevalence in children with Down syndrome

- Retrospective cross-sectional study in large cohort of DS patients in Belgium
- 122 patients categorized both by age groups (0-5, 5-12, 12-18) as well as parental report of sleep-disordered breathing symptoms/signs
  - Overall prevalence of OSA (defined as AHI > 2) was 66.4%
    - Mild: 30%, moderate: 25%, severe: 45%
  - OSA confirmed by PSG in > 50% of patients in all age groups
  - Significant inverse correlation of age with AHI
  - OSA in symptomatic patients: 75.7% (and OSA tended to be more severe)
  - OSA in asymptomatic patients: 53.8% (I)
Predictors of OSA in DS

*Retrospective Study of Obesity in Children with Down Syndrome*

Janet S. Basil, MS\textsuperscript{1,2}, Stephanie L. Santoro, MD\textsuperscript{3,4}, Lisa J. Martin, PhD\textsuperscript{1,2}, Katherine Wusik Healy, MS\textsuperscript{1,2}, Barbara A. Chini, MD\textsuperscript{1,2}, and Howard M. Saal, MD\textsuperscript{1,2}

*(J Pediatr 2016;173:143-8)*

- Large sample of pediatric DS patients with growth measurements and PSG data
- DS significantly more likely to be obese than age matched controls (47.8% vs 12%)
- OSA (AHI > 2) in entire cohort: 74%
  - 85% in obese, 64% in non-obese
  - Moderate-severe OSA (AHI > 5) more likely in obese
Management of OSA in DS: Observation?

- For mild OSA, AHI “normalizes” over 7 months in 46% of children without T&A and in 79% with T&A
  - In obese/overweight, normalization in 29% of non-surgical and in 68% of T&A


- For mild OSA in Down syndrome managed with observation or medication (fluticasone or montelukast):
  - OSA resolved in only 1/13 (7.7%) with observation and 1/10 (10%) with meds
  - Mild OSA persisted in 10/13 (76.9%) with observation and 6/10 (60%) with meds
  - Worsened to moderate or severe in 2/13 (15.4%) with observation and 3/10 (30%) with meds

Management of OSA in DS: PAP?

- Can effectively improve sleep parameters and gas exchange, as well as adverse sequelae of OSA
- However… tolerance and long term adherence is often limited, particularly in those with neurocognitive or behavioral problems
- Some studies report higher rates of adherence with inpatient initiation and modification of pressure settings


Management of OSA in DS: Tonsillectomy & Adenoidectomy?

- Considered first-line treatment of OSA in pediatric patients
- *OSA cure rate of 60-80% of non-syndromic patients


Management of OSA in Down Syndrome: Tonsillectomy & Adenoidectomy?

- Helpful, but high rate of persistent OSA...
  - Merrell & Schott, 2007: ~60% with abnormal post-op PSG; no improvement with addition of lateral pharyngoplasty or pillar plication
  - Shete, 2010: only 18% cure rate
  - Thottam, 2015: Significant improvement in AHI to mild-moderate levels, but only ~5% achieved normal PSG. Majority of those with severe OSA had residual OSA.
  - Abijay, 2021:
    - Complete resolution of OSA in only 5%
    - Mean AHI reduction 27.9 → 14.0; also improvements in arousal index and time O2 < 90%
    - 60% of patients with 50% or more reduction in AHI
    - Persistent OSA associated with age, asthma, and obesity
    - 15.8% with worsening AHI after T&A
Approach to persistent OSA after T&A

- Review sleep study
- Review symptoms
- Room for improvement with medical approach?
  - Poorly controlled environmental allergies
  - Nasal steroid & saline for significant chronic rhinitis
  - Leukotriene receptor antagonist
- Is PAP an option?
Physical Exam:
Sleep apnea is a problem of the upper airway (tip of nose → trachea)

- **General:**
  - voice
  - Stridor
  - BMI
  - Midface and mandibular hypoplasia
  - General tone

- **Nasal:**
  - Structural integrity
  - Septal deviation
  - Mucosal edema
  - Inferior turbinate hypertrophy
  - Polyps/masses
Physical Exam

- Oral cavity:
  - Tonsils
  - Palate
  - Tongue
  - Lingual tonsil (?)
  - Posterior pharyngeal mass
Physical Exam

- Flexible fiberoptic exam
  - Posterior nasal passage
  - Adenoid re-growth
  - Pharynx – asymmetry, collapse, lesions
  - Tongue base – lingual tonsil, masses
  - Larynx – laryngomalacia, masses, cord mobility
Limitations

- Anatomy during office exam ≠ anatomy during sleep
  - Patient position
  - Pharyngeal & laryngeal tone
  - Poor cooperation
State-specific examinations

- Drug-induced sleep endoscopy (DISE)
- Cine MRI
DISE – what are we looking at?

* For each location, assess view obtained at most and least obstructed points in respiratory cycle
Causes of Persistent OSA in DS after T&A

- Common sites include:
  - Tongue (size, position, or both)
  - Lingual tonsil enlargement
  - Collapse of laryngeal structures above the vocal cords (laryngomalacia)
Lingual tonsillectomy
Lingual Tonsillectomy

Meta-analysis (4 studies, 73 patients) of lingual tonsillectomy for persistent OSA in non-syndromic pediatric patients

Mean reduction AHI 8.9 events/hour

Success rates:
- 17% with AHI < 1
- 51% with AHI < 5
- Similar rate/severity of complications compared with palatal tonsillectomy
Lingual tonsillectomy in DS

Polysomnographic Outcomes Following Lingual Tonsillectomy for Persistent Obstructive Sleep Apnea in Down Syndrome

J. Drew Prosser, MD; Sally R. Shott, MD; Oscar Rodriguez, MD; Narong Simakajornboon, MD; Jareen Meinzen-Derr, PhD; Stacey L. Ishman, MD, MPH

- Median AHI 9.1 → 3.7
- Mean oxygen saturation 84% → 89%
- Success rates:
  - 19% with AHI < 1
  - 61.9% with AHI < 5
Midline Posterior Glossectomy
Midline Posterior Glossectomy and Lingual Tonsillectomy in Obese and Nonobese Children With Down Syndrome: Biomarkers for Success

Evan J. Propst, MD, MSc, FRCSC®; Reshma Amin, MD, FRCPC, MSc®; Natasha Talwar; Michele Zaman; Allison Zweerink, NP-Peds; Susan Blaser, MD, FRCPC; Christian Zaarour, MD, BSc; Igor Luginbuehl, MD; Cengiz Karsli, MD, FRCPC; Albert Aziza, MHSc, BHA, MRTR; Christopher Forrest, MD, MSc, FRCSC, FACS; James Drake, MBBCH, FRCSC; Indra Narang, MBBCH, MD

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<th>TABLE II. Polysomnography Results Pre- and Post-MPG With or Without LT.</th>
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<td>Age (yrs), mean (SD) [range]</td>
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<td>Obstructive AHI</td>
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<td>Normal/overweight became obese</td>
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Supraglottoplasty for laryngomalacia

- Systematic review of 4 studies
- Total of 77 patients
- Mean age = 5.7 years
- Mean AHI: pre-op 12.1 → post-op 4.4
- Success:
  - AHI < 5: 58%
  - AHI < 1: 16%

Hypoglossal nerve stimulation (Inspire)

- Strollo, 2014:
  - Significant improvement in AHI and QOL in adults with OSA related to TBO
Hypoglossal nerve stimulation in pediatric DS

- **Diercks, 2016:**
  - 14 year old with DS requiring tracheostomy for persistent OSA
  - AHI 48.5 → 3.4; able to decannulate

- **Yu, 2022:**
  - Multi-center clinical trial of safety and effectiveness for persistent OSA in DS
    - 10-22 years old; AHI > 10
    - Exclusion: central apneas > 25%, BMI > 95%, AHI > 50
  - Results:
    - Mean decrease AHI: 12.9 events/hour
    - 30/41 (73.2%) with AHI < 10
    - 14/41 (34.1%) with AHI < 5
    - 3/41 (7.3%) with AHI < 2
    - Significant improvements in QOL as measured by parental OSA-18 survey
    - Acceptable adverse event profile