

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

Prevalence of Obstructive Sleep Apnea in Children with Down Syndrome

Mieke Maris, MD¹; Stijn Verhulst, MD, PhD²; Marek Wojciechowski, MD³; Paul Van de Heyning, MD, PhD¹; An Boudewyns, MD, PhD¹

SLEEP 2016;39(3):699–704.

- 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% (!)

Predictors of OSA in DS

Retrospective Study of Obesity in Children with Down Syndrome

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

Marcus, et al. A randomized trial of adenotonsillectomy for childhood sleep apnea. New Eng J Med, 2013.

- 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

Howard, et al. Outcomes in children with Down syndrome and mild obstructive sleep apnea treated non-surgically. Laryngoscope, 2019.

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

Verhulst. Long term continuous positive airway pressure and non-invasive ventilation in obstructive sleep apnea in children with obesity and Down syndrome. *Frontiers in Pediatrics*, 2020.

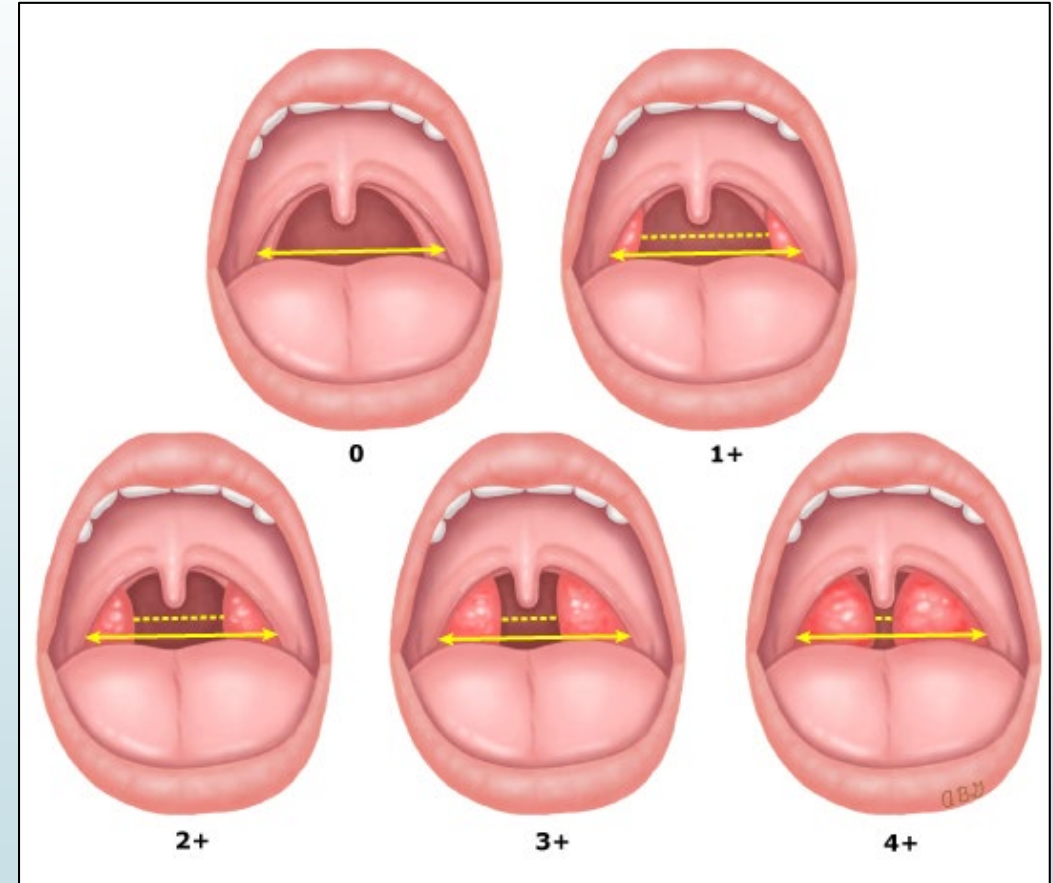
Dudoignon et al. Obstructive sleep apnea in Down syndrome: Benefits of surgery and noninvasive respiratory support. *Am J Med Genet*, 2017.

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

Mitchell, et al. Outcome of adenotonsillectomy for obstructive sleep apnea in obese and normal-weight children. Otolaryngol – Head & Neck Surgery, 2007.

Bhattacharjee, et al. Adenotonsillectomy outcomes in treatment of obstructive sleep apnea in children: a multicenter retrospective study. Am J Resp Crit Care Med, 2010.



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:

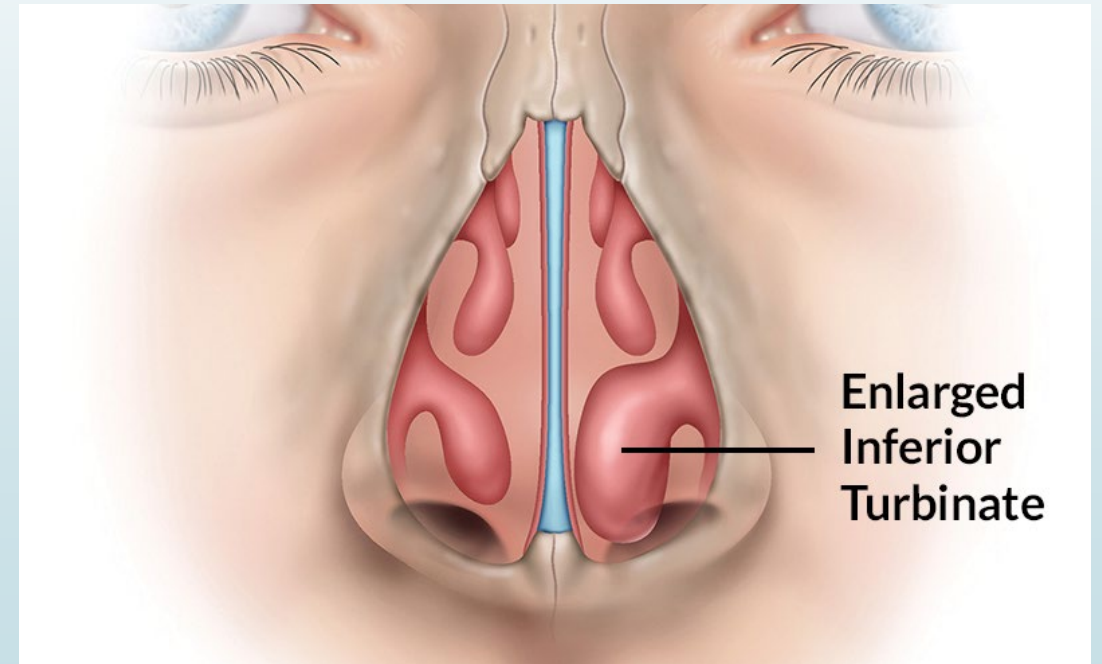
Sleep apnea is problem of the upper airway (tip of nose → trachea)

► General:

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

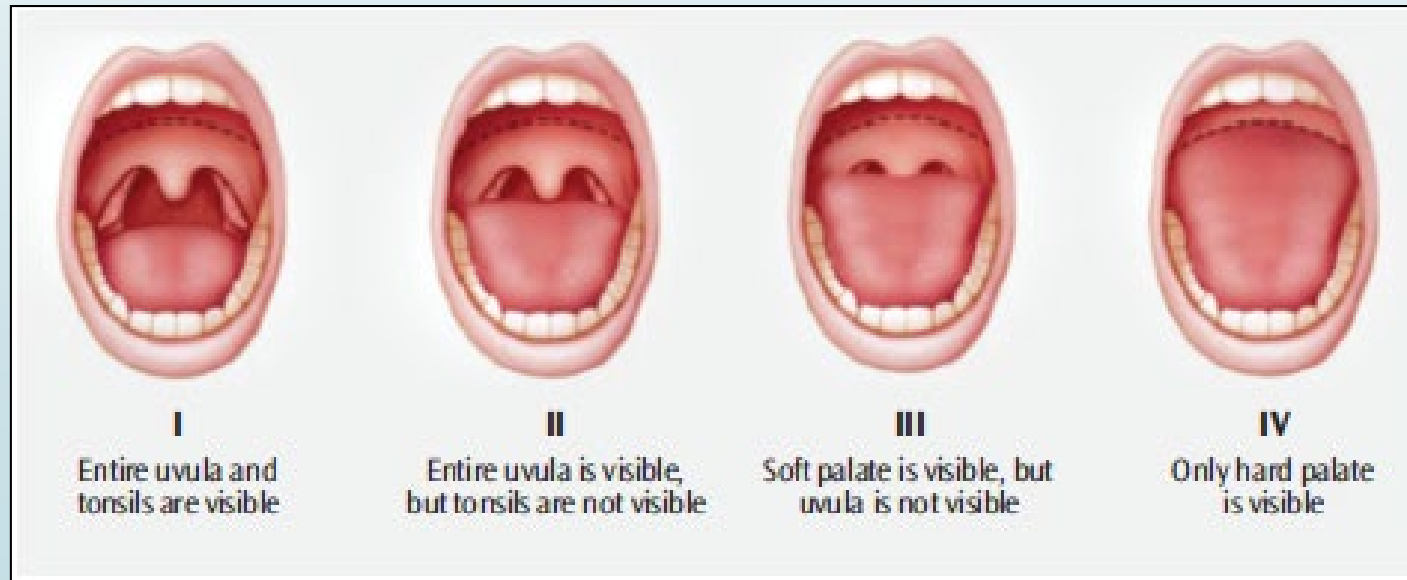
► Nasal:

- Structural integrity
- Septal deviation
- Mucosal edema
- Inferior turbinate hypertrophy
- Polyps/masses



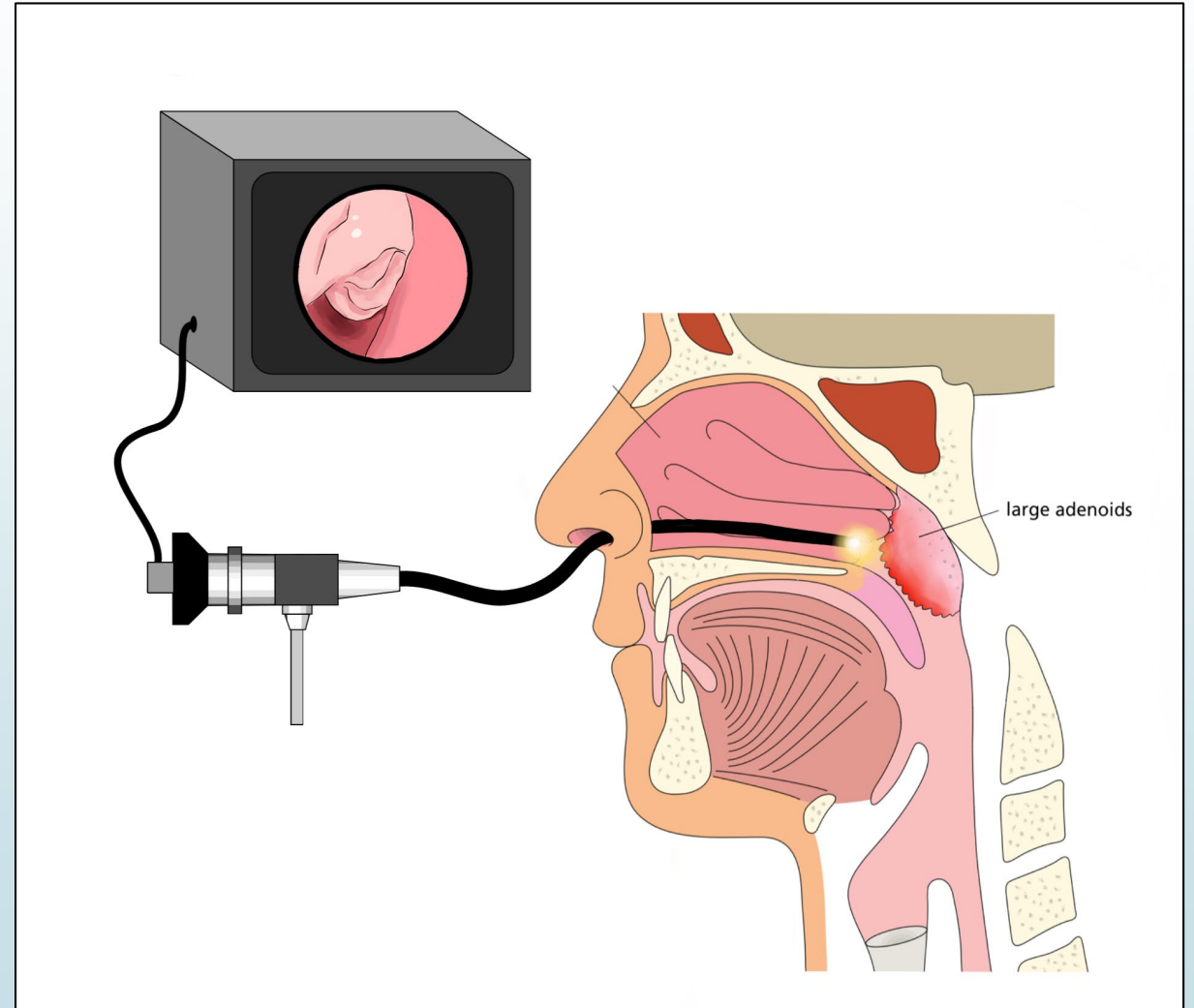
Physical Exam

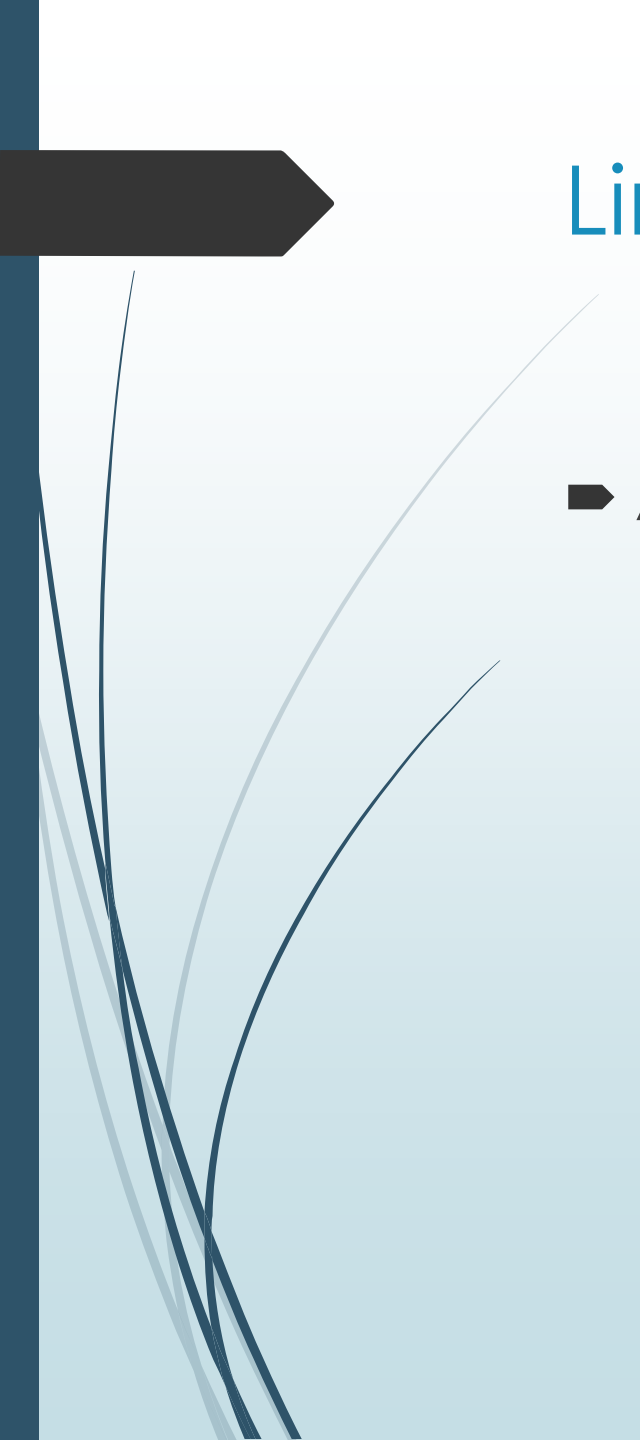
- Oral cavity:
 - Tonsils
 - Palate
 - Tongue
 - Lingual tonsil (?)
 - Posterior pharyngeal mass



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





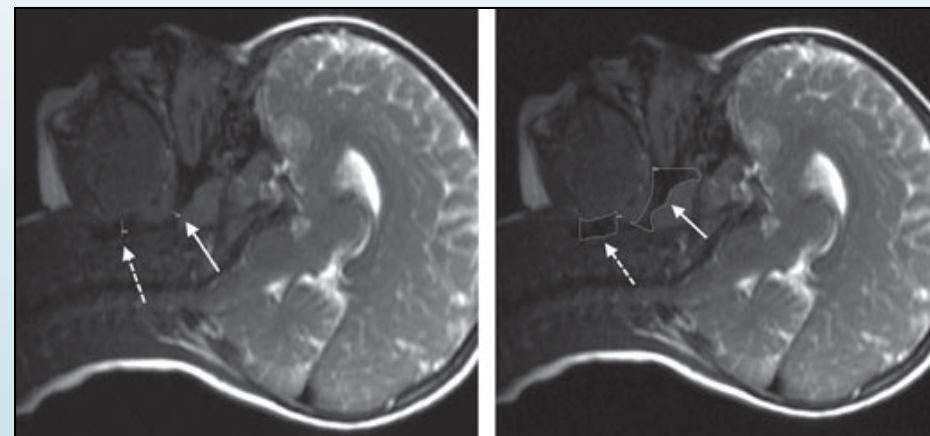
Limitations

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

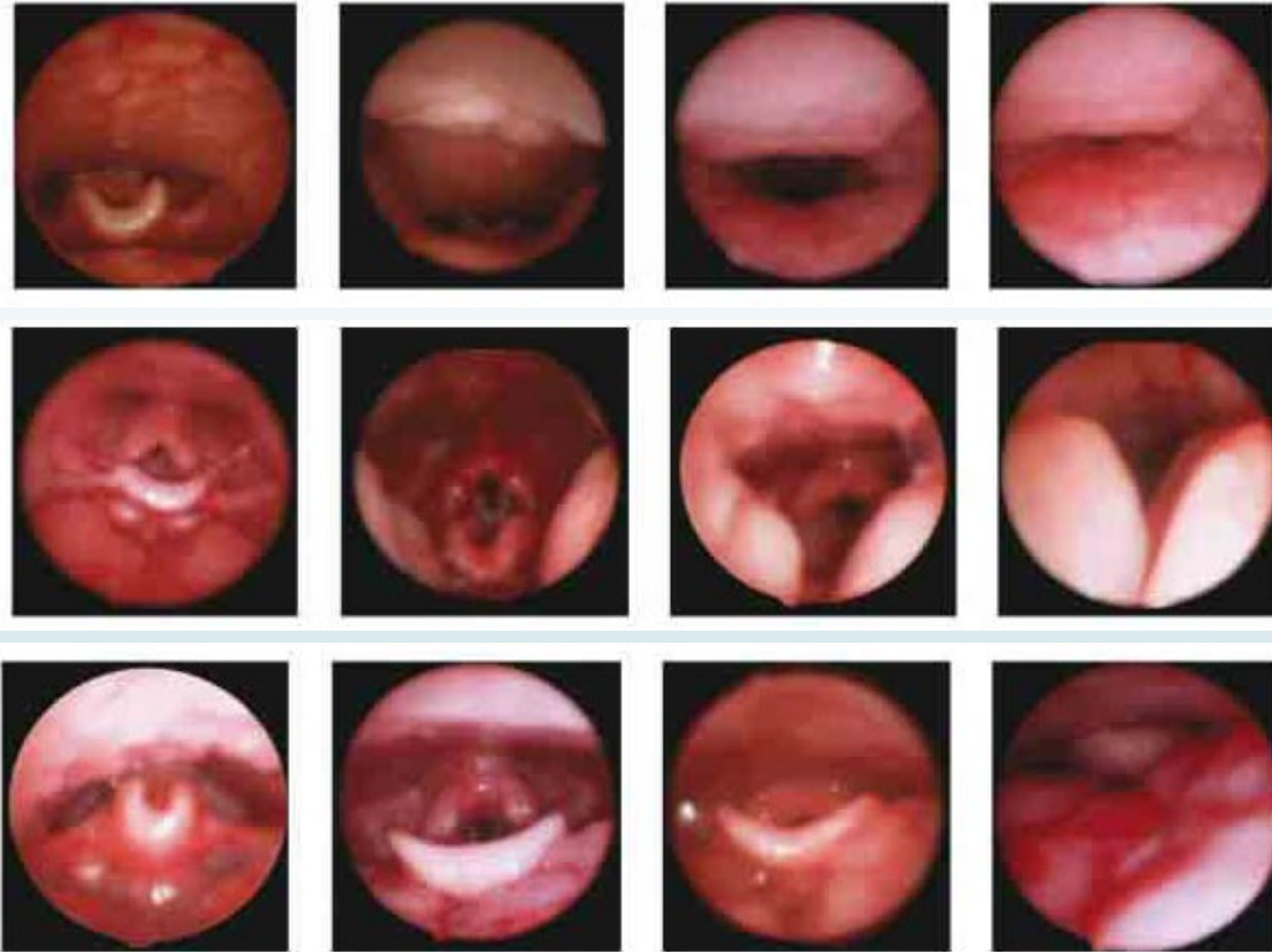
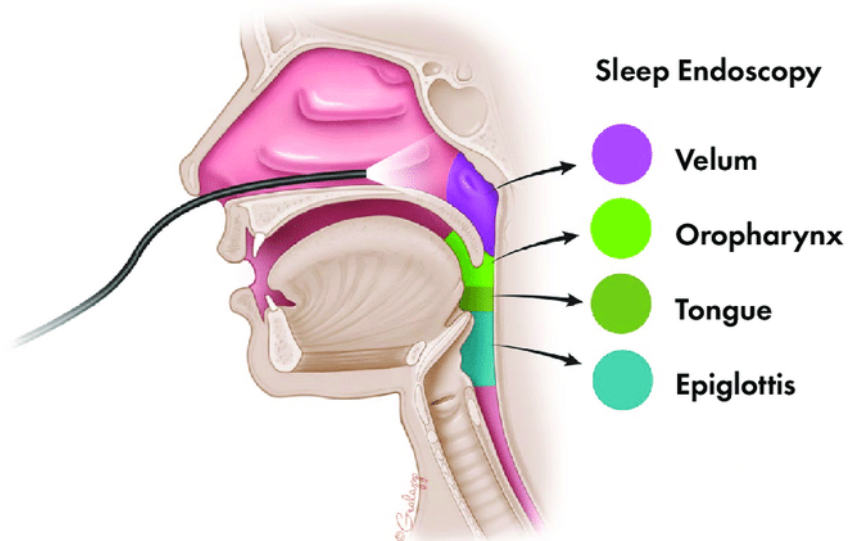
State-specific examinations

► Drug-induced sleep endoscopy (DISE)

► Cine MRI



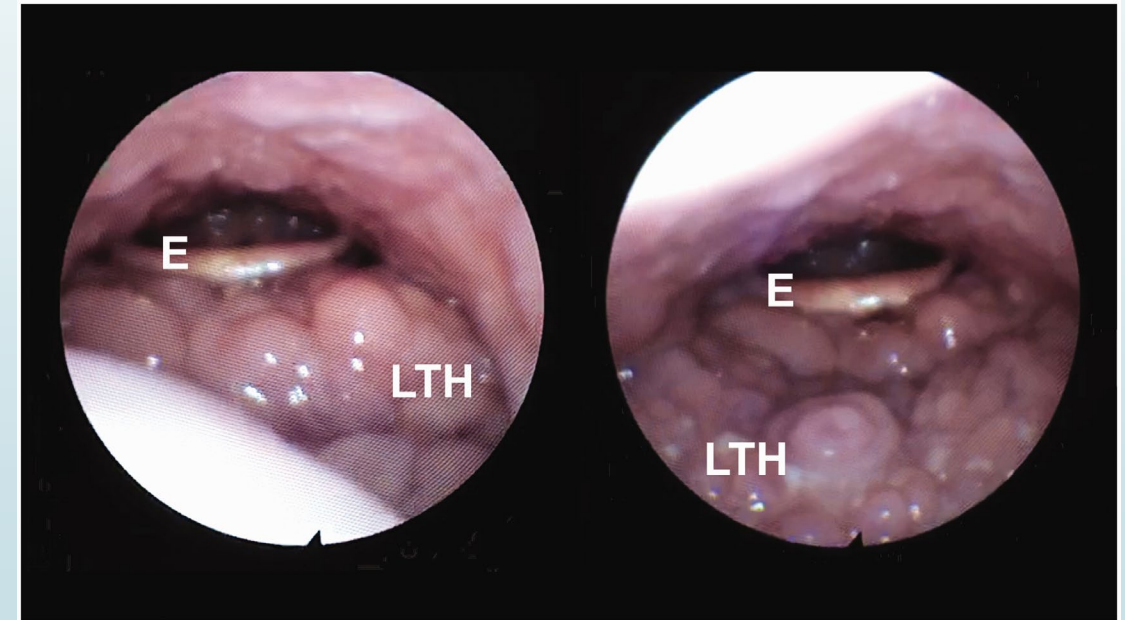
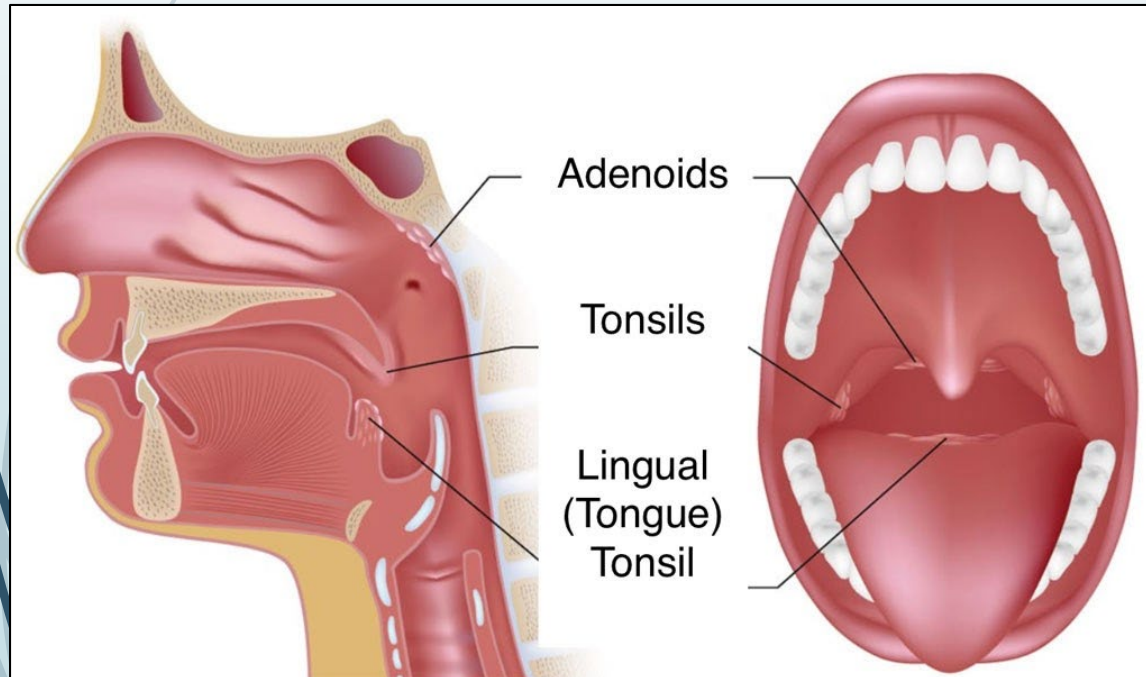
DISE – what are we looking at?



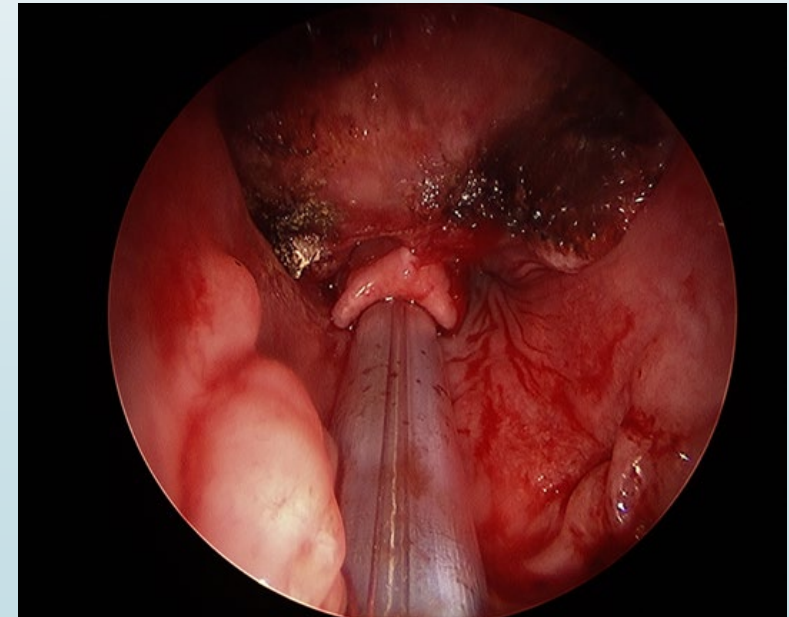
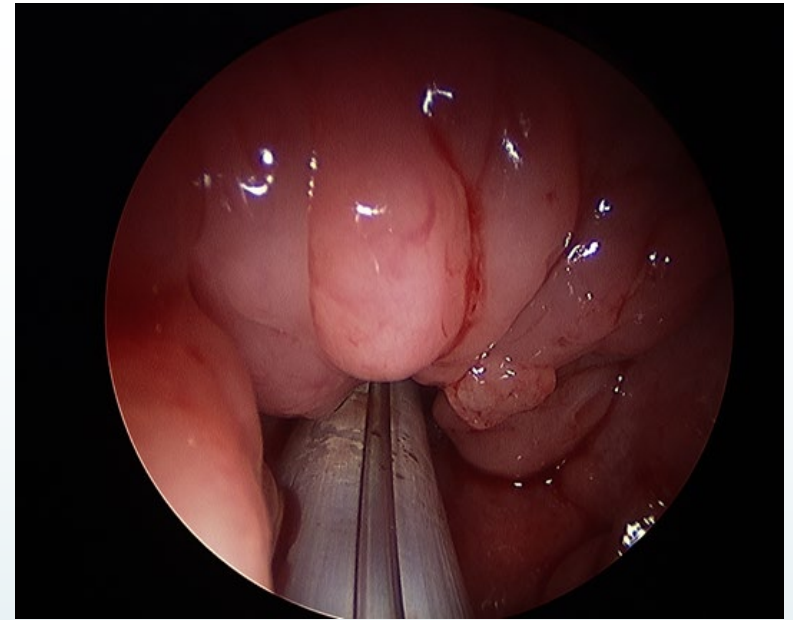
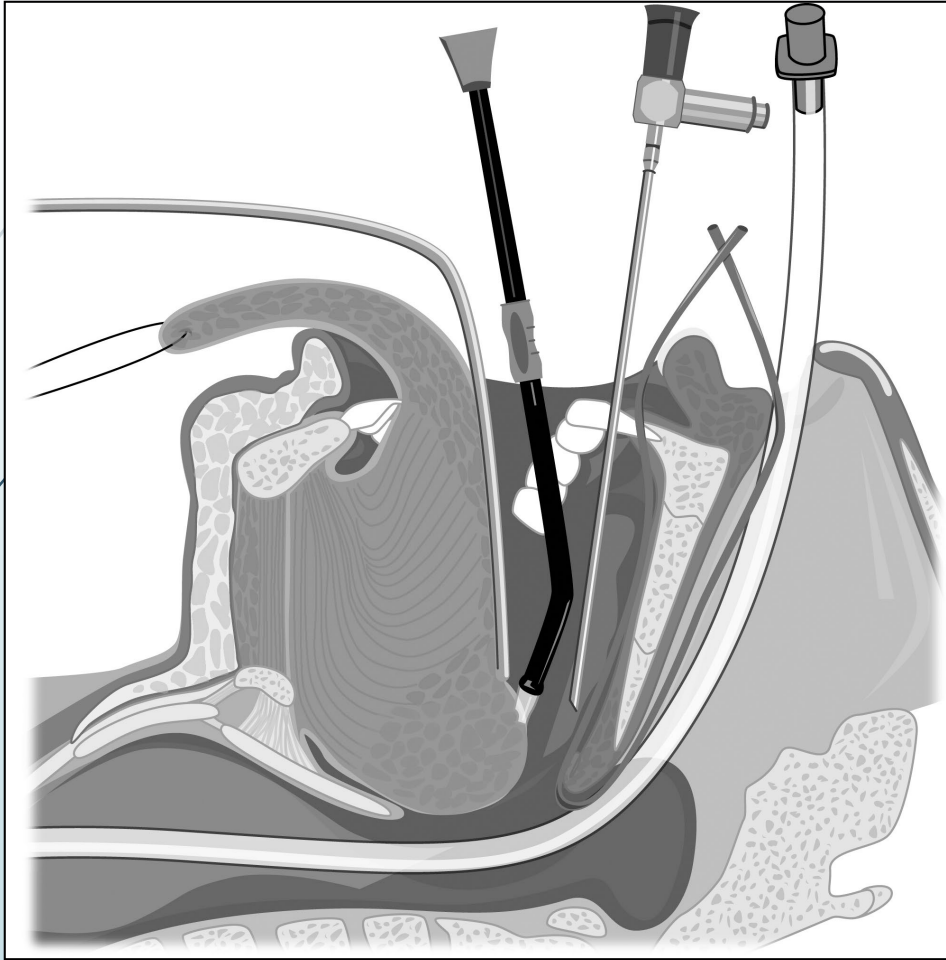
* For each location, assess view obtained at most and least obstructed points in respiratory cycle

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



From: Kille & Lewoczko. Operative Techniques
Otolaryngol. 2015; 26(3): 156-160.

Lingual Tonsillectomy

JAMA Otolaryngology-Head & Neck Surgery | Original Investigation

Lingual Tonsillectomy for Treatment of Pediatric Obstructive Sleep Apnea A Meta-analysis

Kun-Tai Kang, MD, MPH; Peter J. Koltai, MD; Chia-Hsuan Lee, MD, MPH; Ming-Tzer Lin, MD;
Wei-Chung Hsu, MD, PhD

JAMA Otolaryngol Head Neck Surg. 2017;143(6):561-568.

- ▶ 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

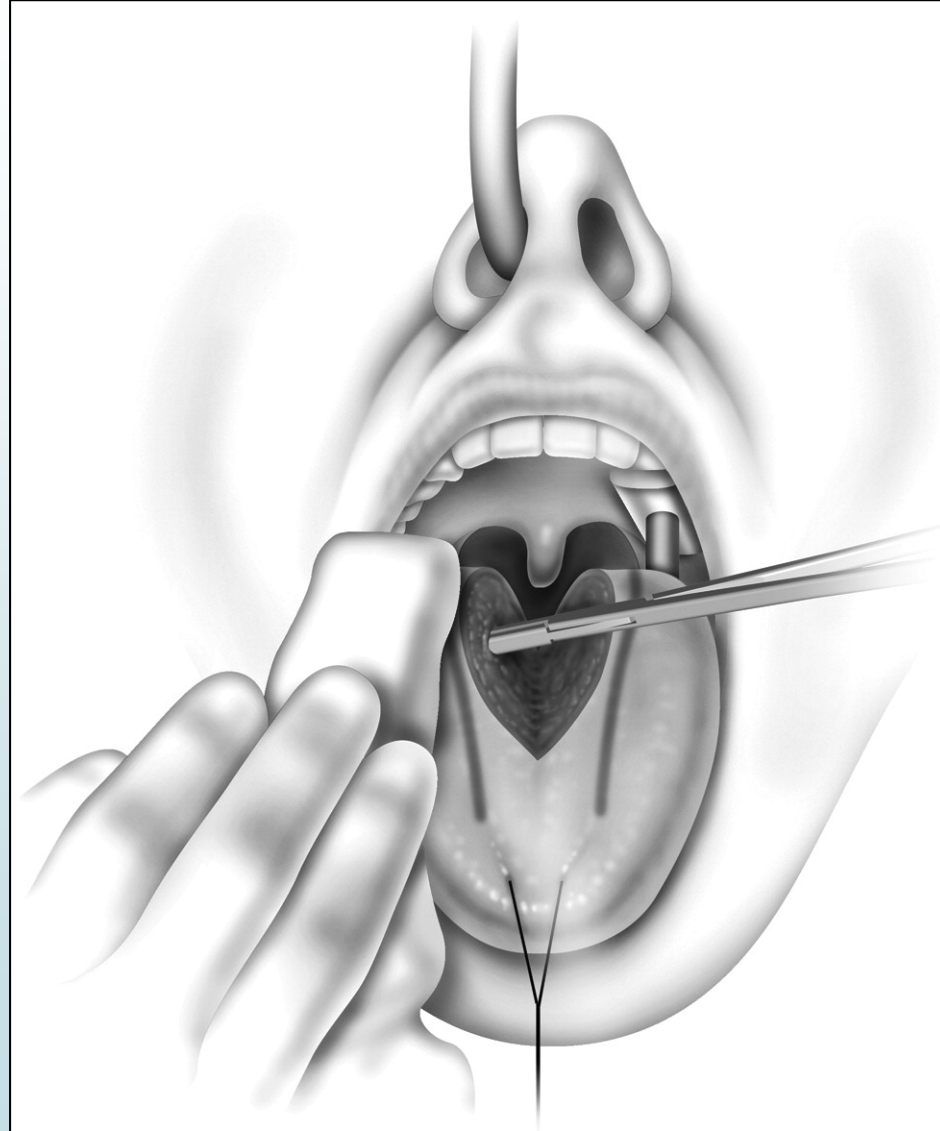
The Laryngoscope
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Rhinological and Otological Society, Inc.

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

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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

TABLE II.
Polysomnography Results Pre- and Post-MPG With or Without LT.

Variable	Preoperative	Postoperative	P Value
Age (yrs), mean (SD) [range]	13.5 (3.9) [6.8–18.7]	14.7 (4.1) [7.5–19.9]	
Time supine (%)	39.4 (43.5) [0–100]	54.2 (43.5) [0–100]	.321
Obstructive AHI	44.3 (30.6) [14.5–114.2]	34.1 (36.4) [9–126.5]	.377
Normal/overweight remained normal/overweight	47.0 (12.8) [37.1–61.4]	5.6 (4.9) [9–10.7]	.031
Normal/overweight became obese	89.8 (25.2) [63.9–114.2]	71.5 (61.7) [4.8–126.5]	.729
Obese remained obese	23.7 (8.6) [14.5–40.5]	30.2 (15.9) [9.1–49.2]	.337

Supraglottoplasty for laryngomalacia

Laryngomalacia

Pre-supraglottoplasty



Post-supraglottoplasty

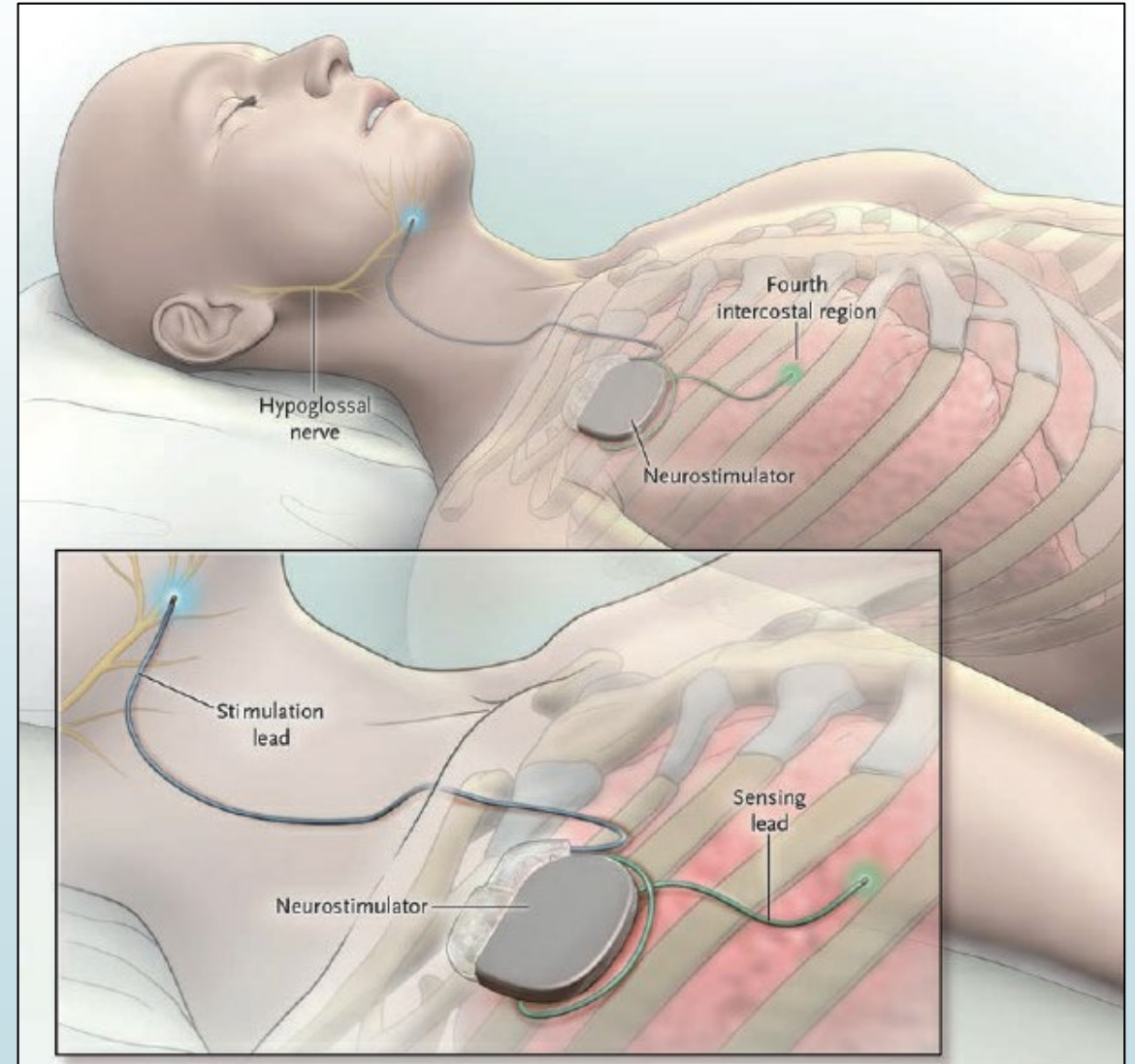


- Systematic review of 4 studies
 - Total of 77 patients
 - Mean age = 5.7 years
 - Mean AHI: pre-op 12.1 → post-op 4.4
 - Success:
 - AHI < 5: 58%
 - AHI < 1: 16%

Manickam, et al. Laryngoscope. 2016;126:491-500.

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