

Evaluation and Management of Obstructive Sleep Apnea in Children with Trisomy 21

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Talk objectives

As a result of participation in this activity, participants will be able to:

- Describe why children with Down syndrome (DS) are at higher risk for obstructive sleep apnea (OSA).
- Identify the common nighttime and daytime signs and symptoms of OSA.
- Define the role of in-lab polysomnography and the potential role of home sleep testing evaluation of OSA in children with DS.
- Cite common treatments and newer potential treatment options for persistent or refractory OSA in children with DS.
- Ask appropriate questions to care teams to demonstrate why repeat evaluation are often necessary.

Why this matters

OSA is far more common in children with Down syndrome than in the general pediatric population.

Reported prevalence in pediatric Down syndrome cohorts/reviews is often about 35–90%, depending on age and definitions.

Untreated OSA can worsen behavior, attention, learning, growth, cardiopulmonary health, and quality of life.

Examples from the attached papers

Cielo 2023: estimated 54–90% prevalence in youth with DS.

Gastelum 2021 review: 66.4% in one large cohort; 76% in a meta-analysis.

Borrelli 2023: OSA found in 67% of their prospective DS cohort.

Cielo et al., J Clin Sleep Med 2023; Gastelum et al., Children 2021; Borrelli et al., J Pers Med 2023.

Why children with Down syndrome are at higher risk

Smaller upper airway and midface/mandibular hypoplasia

Relatively larger tongue (macroglossia) and glossoptosis

Adenoid and tonsil enlargement

Generalized hypotonia, making the airway more collapsible during sleep

Higher rates of obesity, hypothyroidism, reflux, and airway anomalies

Bottom line

Several risk factors can be present at the same time.

That is why OSA can happen even when symptoms seem mild.

Gastelum et al., Children 2021; Cielo et al., J Clin Sleep Med 2023; Hanna et al., Eur Respir Rev 2022.

Common nighttime and daytime signs

Nighttime: snoring, pauses in breathing, gasping, restless sleep, mouth breathing, unusual sleep positions

Daytime: sleepiness, irritability, hyperactivity, attention or learning concerns, morning headaches

Feeding problems, aspiration, laryngomalacia, pulmonary hypertension, and failure to thrive may point to more severe disease in some children

Infants and younger children

Symptoms may be subtle.

Some babies and toddlers have severe OSA without classic snoring histories.

Seither et al., Pediatrics 2023; Gupta & Sharma, Cureus 2024; Gastelum et al., Children 2021.

Symptoms alone are not enough

Parent questionnaires and symptom reports are helpful, but they do not reliably rule OSA in or out.

In one multidisciplinary pediatric DS study, 96% of families reported sleep problems, but questionnaire scores did not correlate well with objective sleep-study results.

This is why many experts recommend objective sleep testing even when symptoms are mild or hard to interpret.

Clinical message

A child can look 'not that sleepy' and still have significant OSA.

Normal-looking nights at home do not always mean a normal sleep study.

Borrelli et al., J Pers Med 2023; Hanna et al., Eur Respir Rev 2022.

In-lab polysomnography (PSG): the reference test

PSG is the gold standard test for diagnosing pediatric OSA.

It measures breathing, oxygen, sleep stages, airflow, effort, and often carbon dioxide.

AAP guidance recommends at least one PSG by age 4 years in children with Down syndrome, with earlier or repeat testing when symptoms or risk factors warrant it.

Why families are often referred

High prevalence of OSA
Symptoms may be misleading
Results can guide surgery, PAP therapy, and follow-up

Knollman et al., Otolaryngol Head Neck Surg 2019; Seither et al., Pediatrics 2023; Hanna et al., Eur Respir Rev 2022.

What the screening studies show

Knollman 2019: among 766 children with DS, 61% underwent PSG and the mean age at first PSG was 4.2 years; only 44.5% had first PSG by age 4.

After the AAP guidance, children were screened at younger ages, but many still were not tested on time.

Seither 2023: among children with documented PSG, 94% had an abnormal study and 60% had moderate or severe OSA.

Take-home point

Guidelines help, but real-world screening still lags.
High-risk infants and young children may need evaluation even earlier.

Knollman et al., 2019; Seither et al., 2023.

Who is most likely to have OSA?

There is no single symptom or exam finding that predicts OSA well.

A 2022 systematic review/meta-analysis found that older age was associated with OSA, but most other predictors were inconsistent.

This supports routine screening plus repeat or longitudinal testing as children grow.

Helpful clues, not perfect rules

Obesity can increase risk.

Airway crowding, larger adenoids/tonsils, and some craniofacial findings may add risk.

Hanna et al., Eur Respir Rev 2022; Borrelli et al., J Pers Med 2023.

Potential role of home sleep testing

Home sleep apnea testing (HSAT) may improve access for families who struggle with lab-based studies.

In youth with Down syndrome, level II HSAT was feasible, often preferred by families, and showed good accuracy for moderate–severe OSA.

But HSAT is not yet a full replacement for in-lab PSG in all children.

Key numbers from Cielo 2023

73.7% of parents preferred HSAT
Sensitivity 0.81; specificity 0.75;
accuracy 0.80
HSAT could expand access in
selected patients

Cielo et al., J Clin Sleep Med 2023.

What home testing can and cannot do

Home studies can miss details captured in the sleep lab, especially when carbon dioxide monitoring or full sleep staging is limited.

General pediatric HSAT data show promise for moderate–severe OSA, but evidence is still developing.

An ongoing randomized trial is studying HSAT versus PSG in children, including those with Down syndrome.

Best current framing for parents

HSAT may be a useful access tool. PSG remains the standard when results are unclear, severe disease is suspected, or treatment decisions are complex.

Revana et al., J Clin Sleep Med 2022; Roman Rosado et al., BMJ Open 2025 protocol; Cielo et al., 2023.

Common treatment options

Adenotonsillectomy is often the first-line treatment when enlarged tonsils/adenoids are contributing.

Positive airway pressure therapy (CPAP or BiPAP) is a main option for persistent OSA, severe OSA, or children who are not surgical candidates.

Adjuncts may include medical treatment for nasal inflammation/allergy, weight management, and evaluation for hypothyroidism or other contributors.

Why treatment is individualized

Children with DS often have multilevel airway obstruction. More than one treatment may be needed over time.

Gastelum et al., Children 2021; Hudson et al., Front Pediatr 2022.

Why repeat evaluation is often necessary after surgery

Tonsil and adenoid surgery often helps, but it does not always cure OSA in Down syndrome.

Diala 2024 found that among 40 children who had a second postoperative PSG, 13 (32.5%) had moderate/severe OSA.

Across the full cohort, 17 of 57 children (29.8%) progressed to moderate/severe OSA, with a median time to later postoperative PSG of 2.3 years.

Practical message

A child can improve at first and still worsen later.

Persistent snoring or return of symptoms should trigger re-evaluation.

Diala et al., Otolaryngol Head Neck Surg 2024.

CPAP/BiPAP and long-term non-invasive ventilation

A 2022 systematic review identified 543 children with DS using long-term non-invasive ventilation.

Efficacy, feasibility, and adherence were generally comparable to other children, though some children with DS had more difficulty getting started.

Outcome data suggest favorable effects on cardiac function and attention in some studies.

Family counseling point

Starting PAP can take patience and desensitization.

Support from sleep teams and respiratory therapists matters.

Hudson et al., Front Pediatr 2022.

Newer and second-line treatment options

Drug-induced sleep endoscopy (DISE) can help identify where the airway collapses and guide targeted treatment.

Additional surgeries may include lingual tonsil surgery or other airway procedures in selected children.

Hypoglossal nerve stimulation (upper airway stimulation) is an emerging option for carefully selected adolescents with refractory OSA.

Review article summary

Persistent OSA often requires stepwise, multidisciplinary care. Newer therapies are usually considered after first-line options have been tried.

Gastelum et al., Children 2021.

Hypoglossal nerve stimulation: what families should know

The 2018 pilot series in 6 adolescents with Down syndrome showed a 56% to 85% reduction in AHI at 6–12 months, with improved quality-of-life scores.

Long-term follow-up published in 2025 reported response rates of 69.7% at 1 year and 87.9% at about 4 years after implantation.

These are promising results, but this remains a specialized therapy for selected older children/adolescents with persistent OSA.

Selection matters

Usually considered after prior surgery and unsuccessful PAP. Requires specialty evaluation and follow-up.

Diercks et al., JAMA Otolaryngol 2018; Larrow et al., Laryngoscope 2025.

Why a multidisciplinary approach helps

Sleep medicine, ENT, pulmonology, genetics, orthodontics, and primary care may all have a role.

Borrelli 2023 found that children with cross-bite plus grade III adenoids and larger tonsils had higher AHI and ODI than those without this combination.

This is one reason treatment plans are often individualized rather than one-size-fits-all.

Possible team members

Sleep specialist
ENT surgeon
Pulmonologist
Orthodontist/dentist
Primary pediatrician

Borrelli et al., J Pers Med 2023.

Questions parents can ask the care team

Has my child had the right type of sleep study yet, and is it time to repeat it?

If surgery is recommended, how will we check for residual OSA afterward?

Would home testing be reasonable for my child, or is in-lab PSG better?

What features suggest my child may need CPAP/BiPAP or a second-line airway evaluation?

Are weight, thyroid disease, reflux, aspiration, or airway abnormalities making OSA worse?

Important follow-up question

What symptoms or changes should prompt us to call sooner?

Derived from the evidence summarized across Knollman 2019; Seither 2023; Cielo 2023; Diala 2024; Gastelum 2021; Hanna 2022.

Big takeaways and the transition to adolescence/adulthood

OSA in Down syndrome is common, often persistent, and often underrecognized.

Testing matters because symptoms alone are unreliable and risk can change over time.

Treatment can help, but repeat evaluation is often necessary after surgery or as children grow.

Adult Down syndrome data also show that OSA remains common and CPAP can be feasible, reinforcing the need for lifelong awareness.

Adult follow-up article

Landete 2020: among clinically suspected adults who underwent sleep study, all had OSA; CPAP adherence >4 h/day in 79.2% and symptomatic improvement in 58.7%.

Landete et al., Am J Med Genet A 2020.