

Down Syndrome, Sleep and Associated Disorders: Discussion Across the Life Span

By Robyn Woidtke, MSN, RN, RPSGT, CCSH, FAAST

Sleep disorders, as well as sleep problems, are extremely common in individuals with Down syndrome (DS), with many issues presenting at birth and persisting throughout the life span. Ensuring that sleep assessment is included throughout the life continuum for patients with DS is quite important, as sleep issues may contribute to behavior and cognitive issues or good sleep may contribute to better functioning. Knowledge and appreciation about this condition, as well as treatment options related to sleep and associated disorders for the individual and families, are essential for the sleep health professional, as the sleep problems we face as a society are exacerbated in individuals with DS. Similar to the lack of sleep inquiry in the general population, it may be that much more needs to be done to address this significant issue in this population.

Overview of Down Syndrome

According to the U.S. Centers for Disease Control and Prevention (CDC),¹ DS is one of the most common chromosomal disorders and affects about one out of every 700 babies, or about 6,000 babies in the U.S. per year, with about 200,000 living with DS at any given time. There is a higher risk with older maternal age; however, as more deliveries are realized in women younger than 35 years old, the number of DS births are higher in that age group.

Screening may consist of maternal blood sampling and sonograms. They are usually performed for all pregnant women, and diagnosis can be made prenatally at various time points – chorionic villi sampling at 10-12 weeks, amniocentesis at 14-20 weeks and percutaneous umbilical cord sampling at 18 weeks. There are times in which the diagnosis may not be made until after birth. In any instance, the

family will need support and counseling regarding the diagnosis and engagement with social services and other specialists as soon as possible to ensure smooth transitions into childhood and beyond.¹⁻⁴

There are three variants of DS. The most common is trisomy 21, which occurs when there is an additional copy of chromosome 21 (i.e., three instead of two). Translocation occurs in about 3% of individuals – in which chromosome 21 is attached to another – and mosaic, in which some, but not all, of the cells have an extra chromosome 21. Mosaicism is the least common of the variants, and these individuals may have less disability than those with the other types. The average life span of individuals with DS has increased over time from about 25-30 years of age to around 60 years of age.¹⁻⁴

Comorbidity and Chronic Illness

There are higher levels of a variety of comorbidities in people with DS than in the general population. These include congenital heart defects, pulmonary hypertension, diabetes and thyroid dysfunction, vision and hearing deficits, gastroesophageal reflux, and sleep disorders.¹⁻⁵ Cognitive ability may also be impacted by associated comorbidities in many individuals, such as comorbid autism in adults with DS.⁶

One of the most common problems, particularly in children, is a poor immune system response.⁷ As we know, poor sleep/sleep deprivation from any cause may exacerbate a compromised immune system. Poor immune response contributes to frequent colds and

The prevalence of sleep disorders is very high in people with DS, including behavioral sleep issues, insomnia and obstructive sleep apnea (OSA).

congestion, potentially leading to pneumonia. Respiratory infections and complications can impact not only the family, but also affect the health care system, as patients with DS who need to be hospitalized usually take longer periods of time to recover and may experience ongoing morbidity and mortality.⁸ Thyroid dysfunction and obesity may also contribute to sleep issues, especially since the obesity rate in individuals with DS is significantly higher than in typically developing (TD) children, although overall obesity is on the rise.¹⁻⁴

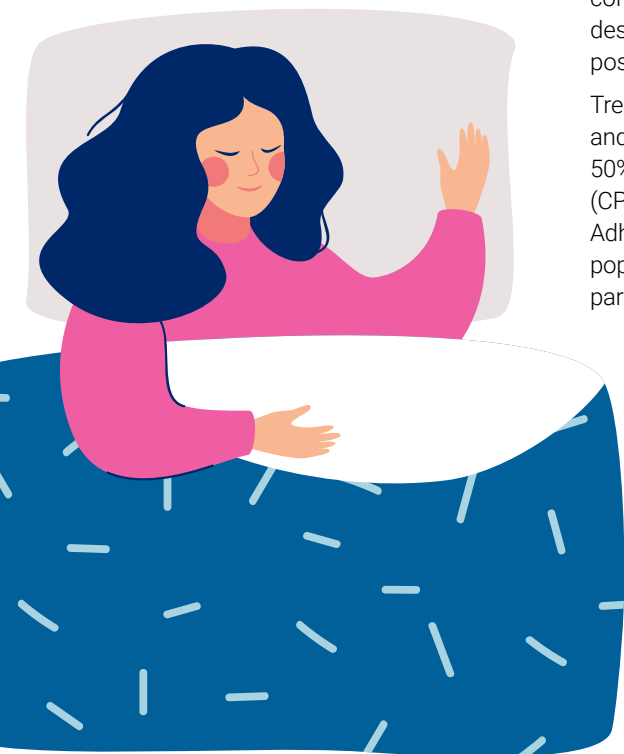
Not limited to medical conditions, age-related mental health issues may arise. According to the National Down Syndrome Society,⁶ in early childhood, these can manifest as anxiety, oppositional behavioral issues, rumination, disruptive activity and hyperactivity. In adolescents and young adults, depression, anxiety and obsessive-compulsive behavior

may be exhibited, as well as social withdrawal. Adults with DS can also experience mental health issues such as anxiety, social withdrawal and dementia, and have a higher incidence of Alzheimer's disease.⁹ Mental health issues may contribute to poor sleep and, as such, they should be addressed and treated.

In a literature review by Gandy et al.,¹⁰ researchers found that of the comorbid conditions they researched — sleep disorders, cardiopulmonary, thyroid and seizures — sleep disorders ranked highest in contributing to cognitive impairment in individuals with DS. Improving sleep, therefore, may be a modifiable factor in enhancing patient outcomes.⁵

Focus on Sleep

The prevalence of sleep disorders is very high in people with DS, including behavioral sleep issues, insomnia and obstructive sleep apnea (OSA). OSA has a significantly higher prevalence in people with DS, compared to the general population, estimated to be 31%-71% compared to 1%-5% in children. There are some estimates as high as 100% in adults.^{11, 12} Reasons for the increased prevalence of OSA can be attributed to multiple features of DS including hypotonia, mid-face hypoplasia, tracheal issues and macroglossia, which



Treatment options for individuals with DS are similar to those of the general OSA population and consist of adenotonsillectomy as first-line therapy in children.

can predispose the individual with DS for OSA.¹¹⁻¹³ Obesity is known to adversely affect sleep and contribute to OSA as well.¹¹ The added burden of comorbidity from OSA is also present, which can create complicated care regimens.

The 2011 American Academy of Pediatrics Clinical Report — Health Supervision for Children with Down Syndrome¹ is quite specific in its recommendations for regular intervals of assessment of sleep and associated disorders, in particular OSA, throughout the child's life, beginning within the first six months of birth and onward with a polysomnogram (PSG) by age 4. However, a 2016 article by Esbensen et al.¹⁴ found that only 47% of patients had a PSG which presents a clinical practice gap. (A 2019 article by Knollman¹⁵ found after the 2011 guideline was published, more PSGs were conducted at an earlier age.) Additionally, a study in Canada¹⁶ found that OSA is persistent throughout childhood; however, repeat PSG was not often performed and, if performed, there was a higher incidence of OSA in the population. In addition, in three out of four individuals, OSA persisted following tonsillectomy and adenoidectomy, furthering the need for ongoing longitudinal sleep assessments.

In some studies, tonsillar hypertrophy and body mass index (BMI) were not associated with an increase of OSA in children. This seems counterintuitive¹¹ but, when taken into the context of the multifactorial inputs, which may be additional causes such as those noted above, it is wise to assess and include other potential sources that may have a contribution to OSA. Additionally found on PSGs were higher amounts of hypoventilation, desaturation and central sleep apnea,⁹ therefore, it is important to use PSG whenever possible and ensure monitoring of CO₂ is included.

Treatment options for individuals with DS are similar to those of the general OSA population and consist of adenotonsillectomy as first-line therapy in children. However, in approximately 50% of patients that present with persistent OSA, continuous positive airway pressure (CPAP), rapid mandibular expansion and other surgical techniques are often used.¹⁷⁻¹⁸ Adherence to CPAP by people with DS is quite similar, or may be better than, the general population. Hill et al.¹⁸ studied adult patients with DS and the use of CPAP. Although the participants had overall low adherence, there were improved outcomes in sleepiness, behavioral and emotional outcomes, and cognition. In a retrospective by Kang et al.¹⁹ of patients with developmental disability (DD) compared to individuals who are TD, those with a DD demonstrated better adherence than the TD. However, the literature is somewhat limited and mixed in this regard, and more studies need to be done to assess CPAP use and interventions to enhance use, therefore alluding to the potential to improve outcomes.

Sleep, Cognitive Function and Behavioral Issues

Sleep and sleep problems greatly contribute to not only cognitive deficits but also to daytime behavioral issues. As we know, a lack of sleep creates a whole host of physiologic dysfunction in individuals that are TD, including reduced immune response and declines in executive function and working memory. Because individuals with DS have a longer time frame to reach developmental milestones,

the importance of adequate sleep is increasingly important.

Sleep disruption is known to adversely impact both cognitive and behavioral issues. Through education and use of techniques to improve sleep, the sleep health community can help parents and DS patients to better address these issues.²⁰

Lastly, do not forget the parents and caregivers. Their sleep, health and well-being may also be impacted. In the early years, the caregivers may have to address a multitude of issues regarding comorbidity, find adequate resources for support and attend to many provider visits for various issues. The stress of immediate care, as well as the knowledge of the long-term issues, may place significant strain on families. In the midst of navigating health care, sleep health is often forgotten.

On a personal note, individuals with DS are a blessing, a source of joy and wonder, and provide us with a unique perspective on life. My niece has DS. She just graduated from high school. The world is her oyster, but she is our pearl. 🌙



**ROBYN
WOIDTKE, MSN,
RN, RPSGT,
CCSH, FAAS,**
*became enamored
by the field of sleep
in 1985. She began
her career in sleep*

in the neonatal field before transitioning into sleep clinic administration roles and eventually the medical sleep device industry. Woidtke holds a master's degree in nursing science and a bachelor's in clinical research administration. She has authored numerous publications and lectured on a variety of sleep-related topics and is currently the principal at Sleep for Nurses, a sleep-focused educational endeavor specifically developed for practicing nurses.

Down Syndrome Resource

[ndss.org/resources/obstructive-sleep-apnea-syndrome](https://www.ndss.org/resources/obstructive-sleep-apnea-syndrome)

References

1. Centers for Disease Control (2021) Facts about Down Syndrome, retrieved June 10, 2021 from <https://www.cdc.gov/ncbddd/birthdefects/downsyndrome.html>
2. Bull, M.J, and the Committee on Genetics Clinical Report: Health Supervision for Children With Down Syndrome. *Pediatrics*. 2011;128(2):393-406. <http://pediatrics.aappublications.org/content/128/2/393>. Reaffirmed January 2018
3. National Association of Down Syndrome (2021) Facts about Down Syndrome, retrieved June 10, 2021 from <https://www.nads.org/resources/facts-about-down-syndrome/>
4. Bull, M. J. (2020). Down syndrome. *New England Journal of Medicine*, 382(24), 2344-2352.
5. Hoffmire CA; Magyar CI; Connolly HV; Fernandez ID; van Wijngaarden E. High prevalence of sleep disorders and associated comorbidities in a community sample of children with down syndrome. *J Clin Sleep Med*. 2014;10(4):411-419.
6. National Down Syndrome Society (2021). Mental Health Issues and Down Syndrome, retrieved June 14, 2021 from <https://www.ndss.org/resources/mental-health-issues-syndrome/>
7. Huggard D, Doherty DG and Molloy EJ (2020) Immune Dysregulation in Children With Down Syndrome. *Front. Pediatr*. 8:73. doi: 10.3389/fped.2020.00073
8. Santoro, S. L., Chicoine, B., Jasien, J. M., Kim, J. L., Stephens, M., Bulova, P., & Capone, G. (2021). Pneumonia and respiratory infections in Down syndrome: A scoping review of the literature. *American Journal of Medical Genetics Part A*. 185(1), 286-299.
9. National Down Syndrome Society (2021) Retrieved June 17, 2021 from <https://www.ndss.org/resources/alzheimers/>
10. Gandy KC, Castillo HA, Ouellette L, Castillo J, Lupo PJ, Jacola LM, et al. (2020) The relationship between chronic health conditions and cognitive deficits in children, adolescents, and young adults with down syndrome: A systematic review. *PLoS ONE* 15(9): e0239040. <https://doi.org/10.1371/journal.pone.0239040>
11. Simpson, R., Oyekan, A. A., Ehsan, Z., & Ingram, D. G. (2018). Obstructive sleep apnea in patients with Down syndrome: Current perspectives. *Nature and science of sleep*. 10, 287.
12. Trois MS; Capone GT; Lutz JA; Melendres MC; Schwartz AR; Collop NA; Marcus CL. Obstructive Sleep Apnea in Adults with Down Syndrome. *J Clin Sleep Med*. 2009 ;5(4):317-323.
13. Nehme, J., LaBerge, R., Pothos, M., Barrowman, N., Hoey, L., Kukko, M., ... & Katz, S. L. (2019). Treatment and persistence/recurrence of sleep-disordered breathing in children with Down syndrome. *Pediatric pulmonology*. 54(8), 1291-1296.
14. Esbensen, A. J., Beebe, D. W., Byars, K. C., & Hoffman, E. K. (2016). Use of sleep evaluations and treatments in children with Down syndrome. *Journal of developmental and behavioral pediatrics: JDBP*. 37(8), 629.
15. Knollman, P. D., Heubi, C. H., Meinzen-Derr, J., Smith, D. F., Shott, S. R., Wiley, S., & Ishman, S. L. (2019). Adherence to guidelines for screening polysomnography in children with Down syndrome. *Otolaryngology–Head and Neck Surgery*. 161(1), 157-163.
16. Ingram DG, Ruiz AG, Gao D, Friedman NR. Success of tonsillectomy for obstructive sleep apnea in children with Down syndrome. *J Clin Sleep Med*. 2017;13(8):975–980.
17. Dudoignon, B., Amaddeo, A., Frapin, A., Thierry, B., de Sanctis, L., Arroyo, J. O., & Fauroux, B. (2017). Obstructive sleep apnea in Down syndrome: Benefits of surgery and noninvasive respiratory support. *American Journal of Medical Genetics Part A*, 173(8), 2074-2080.
18. Hill, E. A., Fairley, D. M., Williams, L. J., Spanò, G., Cooper, S.-A., & Riha, R. L. (2020). Prospective Trial of CPAP in Community-Dwelling Adults with Down Syndrome and Obstructive Sleep Apnea Syndrome. *Brain Sciences*. 10(11), 844. MDPI AG. Retrieved from <http://dx.doi.org/10.3390/brainsci10110844>
19. Kang, E. K., Xanthopoulos, M. S., Kim, J. Y., Arevalo, C., Shults, J., Beck, S. E., ... & Tapia, I. E. (2019). Adherence to positive airway pressure for the treatment of obstructive sleep apnea in children with developmental disabilities. *Journal of Clinical Sleep Medicine*. 15(6), 915-921.
20. Chawla JK, Burgess S, Heussler H. The impact of sleep problems on functional and cognitive outcomes in children with Down syndrome: a review of the literature. *J Clin Sleep Med*. 2020;16(10):1785–1795.