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Sleep-disordered Breathing and Related Comorbidities in Preschool-aged Children with Down syndrome

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Abstract

Sleep-Disordered Breathing (SDB) is prevalent among children with Down syndrome (DS), significantly impacting their health and quality of life. This mini-review explores the prevalence, pathophysiology, clinical manifestations, diagnosis, management, and associated comorbidities of SDB in preschool-aged children with DS. Understanding these aspects is crucial for optimizing care and improving outcomes for this vulnerable population.

Keywords: Down syndrome • Pathophysiology • Breathing

Introduction

Down Syndrome (DS), or trisomy 21, is the most common chromosomal disorder, characterized by a variety of phenotypic manifestations including intellectual disability, congenital heart defects, and distinctive facial features. Among the myriad of health issues faced by individuals with DS. sleepdisordered breathing is particularly significant due to its prevalence and impact on overall health and development. SDB encompasses a spectrum of respiratory problems during sleep, from primary snoring to obstructive sleep apnea, characterized by repeated episodes of partial or complete upper airway obstruction. The prevalence of SDB in children with DS is markedly higher than in the general pediatric population.

Literature Review

Studies estimate that 50-79% of children with DS experience some form of SDB, compared to 1-4% in typically developing children. This increased prevalence is attributed to anatomical and physiological factors inherent in DS, including hypotonia, midfacial hypoplasia, and adenotonsillar hypertrophy. Anatomical Factors: Children with DS often have craniofacial abnormalities such as midface hypoplasia, a narrow nasopharynx, and a relatively large tongue, all of which contribute to airway obstruction during sleep. Adenotonsillar hypertrophy is also more common, exacerbating the risk of airway blockage. Hypotonia, or reduced muscle tone, is a hallmark of DS and affects the muscles of the upper airway, making them more prone to collapse during sleep. This is compounded by generalized muscle weakness affecting the respiratory muscles [1].

Central nervous system abnormalities in DS can impair the neural control of breathing. This can lead to central sleep apnea, characterized by disrupted signals from the brain to the respiratory muscles. Obesity, prevalent among children with DS, further increases the risk of SDB due to the deposition of adipose tissue around the upper airway, reducing its patency. These include

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snoring, observed apneas, restless sleep, frequent awakenings, sweating, and enuresis (bedwetting). Parents may also report choking or gasping during sleep. Daytime manifestations can include excessive sleepiness, behavioral problems, hyperactivity, difficulty concentrating, and morning headaches. Poor sleep quality can also exacerbate cognitive impairments and affect overall development [2].

Discussion

SDB is associated with cardiovascular complications such as hypertension, pulmonary hypertension, and left ventricular hypertrophy. These are particularly concerning in children with DS, who are already at an increased risk for congenital heart defects. A thorough history and physical examination are essential. Clinicians should inquire about nighttime and daytime symptoms, as well as perform a detailed examination of the upper airway. Tools like the Pediatric Sleep Questionnaire (PSQ) can help in screening for SDB. However, they are not diagnostic. The gold standard for diagnosing SDB is an overnight sleep study or polysomnography. PSG provides comprehensive data on sleep architecture, respiratory events, oxygen saturation, and heart rate. It is particularly important for distinguishing between different types of SDB, such as obstructive versus central sleep apnea [3].

Additional assessments may include oximetry, capnography, and imaging studies like lateral neck X-rays or MRI to evaluate anatomical abnormalities. Weight management through diet and exercise is crucial, especially in obese children. Positional therapy can also help, as SDB can be worse in the supine position. Nasal corticosteroids and leukotriene receptor antagonists may reduce inflammation and adenotonsillar hypertrophy. Adenotonsillectomy is often the first-line surgical treatment for OSA in children with DS. This procedure can significantly reduce the severity of SDB. In some cases, additional surgeries such as uvulopalatopharyngoplasty, tongue reduction, or maxillofacial surgery may be necessary. Continuous positive airway pressure (CPAP) or bilevel positive airway pressure (BiPAP) therapy is highly effective for managing OSA, especially when surgery is contraindicated or insufficient [4].

Adherence to PAP therapy can be challenging, and requires careful titration, patient education, and support. In select cases, dental appliances designed to keep the airway open during sleep may be beneficial. SDB in children with DS is associated with several comorbidities that complicate management and impact overall health. Poor sleep quality can exacerbate cognitive impairments and behavioral problems in children with DS, affecting their learning and social interactions. Untreated SDB can lead to serious cardiovascular issues, including systemic and pulmonary hypertension, which can worsen congenital heart disease outcomes. SDB is linked to insulin

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resistance, glucose intolerance, and dyslipidemia, increasing the risk of metabolic syndrome. SDB can affect growth hormone secretion, leading to growth delays in children with DS.

Children with DS have altered immune function, and SDB can further impair immune responses, increasing susceptibility to infections.Continued research is vital to improve the understanding and management of SDB in children with DS. Investigating the genetic factors that predispose children with DS to SDB could provide insights into targeted therapies. Development of new treatment modalities, including less invasive surgical techniques and improved PAP devices, can enhance management outcomes. Long-term studies are needed to assess the impact of early diagnosis and intervention on the overall health, cognitive development, and quality of life of children with DS and SDB. Collaborative care models involving pediatricians, otolaryngologists, pulmonologists, cardiologists, and sleep specialists can optimize management and improve outcomes for children with DS and SDB [5,6].

Conclusion

Sleep-disordered breathing is a common and significant problem in preschool-aged children with Down syndrome, impacting their health, development, and quality of life. Early recognition, accurate diagnosis, and comprehensive management are essential to mitigate the adverse effects of SDB and improve overall outcomes. Continued research and multidisciplinary care approaches are crucial to advancing the understanding and treatment of this complex condition, ensuring better health and quality of life for children with Down syndrome.

Acknowledgement

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Conflict of Interest

None.

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