

Sleep Apnea

“Sleep apnea is a sleep disorder characterized by pauses in breathing or instances of shallow or infrequent breathing during sleep. Each pause in breathing, called an apnea, can last for several seconds to several minutes, and may occur, by definition, at least 5 times in an hour. Similarly, each abnormally shallow breathing event is called a hypopnea. Sleep apnea is classified as a dyssomnia, meaning abnormal behavior or psychological events occur during sleep. When breathing is paused, carbon dioxide builds up in the bloodstream. Chemoreceptors in the blood stream note the high carbon dioxide levels. The brain is signaled to wake the person sleeping and breathe in air. Breathing normally will restore oxygen levels and the person will fall asleep again. Sleep apnea is often diagnosed with an overnight sleep test called a polysomnogram, or "sleep study".

There are three forms of sleep apnea: central (CSA), obstructive (OSA), and complex or mixed sleep apnea (i.e. a combination of central and obstructive) constituting 0.4%, 84%, and 15% of cases, respectively. In CSA, breathing is interrupted by a lack of respiratory effort; in OSA, breathing is interrupted by a physical block to airflow despite respiratory effort, and snoring is common. According to the National Institutes of Health, 12 million Americans have OSA. There are more cases of sleep apnea still because people either do not report the condition or do not know they have sleep apnea.

Regardless of type, an individual with sleep apnea is rarely aware of having difficulty breathing, even upon awakening. Sleep apnea is recognized as a problem by others witnessing the individual during episodes or is suspected because of its effects on the body. Symptoms may be present for years (or even decades) without identification, during which time the person may become conditioned to the daytime sleepiness and fatigue associated with sleep disturbance. Sleep apnea affects not only adults but some children as well.” https://en.wikipedia.org/wiki/Sleep_apnea

Obstructive Sleep Apnea & Down Syndrome

There is a 50-100% incidence of obstructive sleep apnea in individuals with Down Syndrome, with almost 60% of children with Down syndrome having abnormal sleep studies by age 3.5 - 4 years. The overall incidence of obstructive sleep apnea increases as children grow older.

Why Do Individuals With Down Syndrome Have a Higher Incidence of Obstructive Sleep Apnea? – Anatomy accounts for many of the reasons why there is a higher incidence of obstructive sleep apnea (OSA) in individuals with Down syndrome. Some of those factors include: central apnea, low muscle tone in the mouth and upper airway, poor coordination of airway movements, narrowed air passages in the midface and throat, a relatively large tongue, and hypertrophy (enlargement) of adenoid and tonsillar tissues. Increased upper airway infections and nasal secretions and a higher incidence of obesity further contribute to collapse and obstruction of both the oropharynx and the hypopharynx when the individual is sleeping.

What Are the Effects of Obstructive Sleep Apnea? – Sleep disordered breathing has been shown to affect cognitive abilities, behavior, growth rate and more the more serious consequences of pulmonary hypertension (abnormally high blood pressure in the arteries of the lungs) and cor pulmonale (failure of the right side of the heart). Because of the high incidence of underlying congenital heart problems in individuals with Down syndrome, there is a higher risk of development of the more severe complications. Abnormalities in pulmonary vasculature (the circulatory system in the lungs) also increases risk of development of pulmonary hypertension (abnormally high blood pressure in the arteries of the lungs).

Unfortunately, the ability of parents to predict sleep abnormalities in their children with Down syndrome has been shown to be poor. A sleep study or polysomnogram continues to be the gold standard test from which to evaluate sleep disordered breathing and sleep apnea. Because of the poor correlation between parental reporting and sleep study results, the new American Academy of Pediatrics health care guidelines published in Pediatrics in 2011 recommend a baseline sleep study or polysomnogram for all children with Down syndrome by age four. Sleep

apnea is often undetected in both children and adults, so caregivers should monitor sleep patterns in individuals of all ages, especially if there has been a change in mood, behavior or ability to concentrate.

What Are Symptoms of Sleep Abnormalities? – Symptoms that are suggestive of sleep abnormalities include: restless sleep, snoring, gasping noises, heavy breathing, apneic pauses, frequent waking during the night, trouble getting out of bed, daytime sleepiness and excessive napping. Sleep apnea can also cause behavioral changes, including symptoms of irritability, poor concentration and impaired attention. Uncommon sleep positions such as sleeping sitting up, sleeping with the neck hyper-extended or sleeping bent forward at the waist in a sitting position are all suggestive of a sleep disorder or obstructive sleep apnea.

What Causes Obstructive Sleep Apnea? – Enlargement of the tonsils and adenoids is one of the most common causes of obstructive sleep apnea in children. However, other causes of obstruction such as chronic rhinorrhea and congestion, nasal septal deviation, and nasal turbinate enlargement need to be assessed and treated. If the oral exam shows edema of the posterior pharyngeal wall, thus decreasing the size of the posterior pharyngeal airway, gastro-esophageal reflux (GERD) or chronic post-nasal drainage should be considered. Treatment with anti-reflux medications and/or decongestants, nasal steroid sprays and antihistamines can sometimes be helpful.

What Are the Treatments? – If the primary care physician identifies any airway disturbances during sleep, a referral to an otolaryngologist should be done to determine if a sleep study and/or surgical intervention is needed. Non-invasive treatment options include a continuous positive airway pressure (CPAP) machine and weight loss.


Removal of enlarged tonsils and adenoids is the first line surgical treatment. In individuals with Down syndrome, because of their midface hypoplasia and contracted nasopharynx, even mildly enlarged tonsil and adenoids may have a greater than expected effect in regards to airway obstruction. If the tonsils and adenoids do not appear enlarged, it has been suggested that a sleep study should be done to confirm that the individual does not have sleep apnea.

Although tonsillectomy and adenoidectomy (T&A) is the most common initial surgical intervention, studies have shown that persistent obstructive sleep apnea after T&A is possible and more common in individuals with Down syndrome. Further interventions may be needed, both surgical and medical. All of these studies illustrate the need for post-operative evaluation of patients with Down syndrome for residual sleep apnea after T&A surgery with a post-operative sleep study or polysomnogram. Because of the higher rate of respiratory complications after removal of the tonsils and adenoids in individuals with Down syndrome, overnight observation in the hospital after this surgery is also recommended.

If residual obstruction is present despite T&A surgery, medical treatments such as continuous positive pressure ventilation (CPAP/BiPAP) and oxygen use during sleep are still an option. Weight loss can also help to alleviate symptoms. Evaluations to determine the site or sites of residual airway obstruction include flexible nasopharyngoscopy and laryngoscopy examination in the office to rule out enlarged lingual tonsils, residual or regrowth of adenoid tissue, and possible glossoptosis. Radiographic studies using cine MRI studies have shown that the base of tongue obstruction from a combination of relative macroglossia and glossoptosis, enlarged lingual tonsils and also adenoid regrowth are some of the most common sites of residual obstruction in individuals with Down syndrome despite previous T&A.

Surgical options for persistent obstructive sleep apnea in children with Down syndrome need to be tailored to each child's individual pattern of obstruction. Surgical approaches currently being used include lingual tonsillectomy, uvulopalatopharyngoplasty, midline posterior glossectomy, genioglossus advancement, hyoid advancement and craniofacial surgery, including mandibular and midface advancements. Dental appliances to promote mandibular stabilization have also been shown to be helpful in cases of mild residual sleep apnea. However, in cases of severe sleep apnea with associated pulmonary hypertension, severe hypoxemia and/or cardiac complications, tracheostomy may also need to be considered.

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