

Obstructive Sleep Apnea in Children with Down Syndrome: Why does my child need a sleep study?

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Studies report a 50-100% incidence of obstructive sleep apnea (OSA) in individuals with Down syndrome (DS), with almost 60% of children with DS having abnormal sleep studies by age 3.5-4 years (Shott 2006b). Further evidence shows that these numbers increase as children grow older. (Marcus 1991, Levanon 1999, Dyken 2003). Fitzgerald et al showed a 97% incidence of OSA in children with DS who snored, ages 0.2 to 19 years (4.9 years mean age) (Fitzgerald 2007). Predisposing anatomic factors that are present in children with DS include midface hypoplasia and mandibular hypoplasia causing the upper airway size to be smaller than in other children. In addition, children with DS have a relative enlargement of the tongue, medially displaced tonsils that cause more obstruction in the back of the throat, adenoids sitting in a contracted nasopharynx and thus causing more obstruction, as well as hypotonia of the upper airway with resultant collapse at multiple levels of the airway during sleep. Increased upper airway infections and nasal secretions, obesity and hypotonia further contribute to both oropharyngeal and hypopharyngeal (ie. the lower throat) collapse and obstruction with sleep.

Even mild primary snoring has been shown to affect a child's school performance. The more serious sleep disturbed breathing seen in OSA has been shown to affect cognitive abilities and IQ scores, behavior, growth rate and also has more serious consequences of systemic hypertension, pulmonary hypertension and heart failure (Rowland 1981, Levine 1982, Southall 1987, Marcus 1991 Bonnet, 1989). Because of the high incidence of underlying congenital cardiac anomalies in individuals with DS, there is a higher risk of development of the more severe complications (Jacobs 1997). Congenital abnormalities in the pulmonary vasculature also increases risk of development of pulmonary hypertension.

Unfortunately, the ability of parents to predict sleep abnormalities in their children with DS has been shown to be poor (Shott 2006b, Marcus 1991, Ng 2006). A sleep study or polysomnogram continues to be the gold standard test from which to evaluate sleep disturbed breathing and sleep apnea. Because of the poor correlation between parental reporting and sleep study results, a baseline sleep study or polysomnogram is suggested for all children with DS at age 4 years (Shott SR, Amin R, Chini B, et al. 2006, Ng DK, Chung-hong C 2007). This sleep study recommendation was also added to the American Academy of Pediatric guidelines for healthcare for children with Down Syndrome in 2011 (American Academy of Pediatrics 2011).

The Sleep Study or Polysomnogram

A polysomnogram or sleep study is an overnight study where objective data regarding sleep is collected. Oxygen levels are monitored and sleep behaviors such as sleep pauses or apneas are recorded. Sometimes these pauses during sleep are due to obstructive events and sometimes they are due to central events when the brain stops telling you to breathe. The sleep study can differentiate between these. The sleep study also evaluates the sleep stages to determine if you are getting enough REM sleep, the most restful phase of sleep. Sleep fragmentation can also be determined – some people have repeated arousals throughout the night in response to partial obstructions so they are not getting truly restful sleep. In addition to monitoring oxygen levels, the sleep study also monitors carbon dioxide levels. In normal respirations, you breathe in oxygen and breathe out carbon dioxide. In many children with DS, due to shallow respirations or hypoventilation, the carbon dioxide levels are high. Over time, high carbon dioxide levels can cause systemic hypertension and cardiac rate instability.

Sleep studies in children have different definitions and techniques of analysis compared to adult sleep studies. It is better to go to a pediatric sleep lab if possible because of these differences to insure correct data analysis of sleep study results. In addition, the sleep technicians at pediatric centers are more familiar with doing sleep studies on children who may repeatedly try to remove the multiple monitors that are used in a sleep study. As a parent, you should expect that your child will try to remove the monitors (many times) throughout the night. Stay calm as they can easily be replaced. The monitors do not hurt. The sleep technicians at pediatric sleep labs are trained to be patient and understanding.

Presenting Symptoms

Starting at birth, parents should observe their child for restless sleep, snoring, heavy breathing, uncommon sleep positions, frequent waking during the night, daytime sleepiness, apneic pauses, behavior problems associated with poor sleeping. Sleep positions should also be monitored such as sleeping sitting up, sleeping with the neck hyper-extended, or sleeping bent forward at the waist in a sitting position.

Although the focus of treatment for sleep disturbed breathing tends to center around the tonsils and adenoids, other causes of upper airway obstruction include chronic rhinorrhea and nasal congestion, nasal septal deviation, and nasal turbinate enlargement. If there is edema of the posterior pharyngeal wall, thus decreasing the size of the posterior pharyngeal airway, gastro-esophageal reflux (GER) or chronic post-nasal drainage should be considered as the cause of these findings. Treatment with anti-reflux medications and/or decongestants, nasal steroid sprays and/or antihistamines should be considered. It is also important to focus on a good diet and weight control as well as continued exercise as children grow older. Obesity, especially a large weight gain in a short period of time is a major risk factor for OSA.

If there is any question of airway disturbances during sleep, an examination by an Otolaryngologist should be done to determine if a sleep study and/or surgical intervention is needed. Similar to all children, removal of enlarged tonsils and adenoids (T&A) is the first line surgical treatment. In children with DS, 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 child does not have sleep apnea (Fitzgerald 2007).

Although T&A is the most common initial surgical intervention, studies have shown that residual airway obstruction after this surgery is common in children with DS and further interventions may be needed, both surgical and medical (Merrell 2006, Shott 2006b, Donaldson 1988, Jacobs 1996, Shott and Donnelly 2004). Persistent OSA after T&A surgery has recently also been shown to be more common than previously believed in typical children. Mitchell in 2007 showed a 10-20% incidence of persistent sleep apnea in a group of 79 typical children after T&A. Tauman et al., using a much more strict definition of surgical cure showed complete

normalization of all components evaluated in a sleep study in only 25% of their test population of ‘typical’ children (Tauman 2006). This compares to the 5% total success rate seen in the paper by Shott, Amin et al. where a similarly strict definition of “cure” was used in a group of children with DS (Shott 2006b). If ‘cure’ is more akin to the definitions used in the study by Mitchell, almost 50-70% of the children with DS in this study continued to have OSA after T&A.

All of these studies illustrate the need for post-operative evaluation of children with DS 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 (T&A) in children with DS, overnight observation in the hospital after this surgery is also recommended (Bower 1995).

If residual obstruction is present despite T&A surgery, medical treatment options include continuous positive pressure ventilation (CPAP/BiPAP), weight loss, and oxygen use with sleep. Nasal steroid sprays and some anti-leukotrine medications such as Singulair have been shown to be possibly helpful if the residual OSA is mild. Dental appliances to promote mandibular stabilization have also been shown to be helpful in cases of mild residual sleep apnea and are an option for older children who have all secondary teeth in place. If the sleep apnea is only present when sleeping on one’s back, there are devices to encourage side or stomach position sleeping.

Evaluations to determine the site or sites of residual airway obstruction include flexible nasopharyngoscopy and laryngoscopy exams which can be done with the child awake in the office or in the operating room under general anesthesia with flexible endoscopes. These exams can evaluate for enlarged lingual tonsils, residual or regrowth of adenoids and glossoptosis, where the tongue falls back into the airway during sleep. The endoscopic exam also includes evaluation of the larynx and trachea. Radiographic studies using cine MRI studies have shown that obstruction occurs at the base of the tongue from a combination of relative tongue enlargement or macroglossia and glossoptosis. Enlarged lingual tonsils and also adenoid regrowth are also common sites of residual obstruction seen on MRI studies in individuals with DS despite previous T&A (Donnelly 2004).

Regarding surgical options after T&A, for those with DS, similar to all patients, the ideal surgical approach may be different for each patient and needs to be tailored according to each

child's individual pattern of obstruction. Surgical approaches currently being used include revision adenoidectomy, lingual tonsillectomy, uvulopalatopharyngoplasty and expansion pharyngoplasty surgery to enlarge the posterior oropharynx opening, midline posterior glossectomy to decrease the size of the posterior tongue, genioglossus suspension to keep the tongue from falling back into the airway, hyoid advancement to open up the hypopharynx, and craniofacial surgery, including mandibular and midface advancements. 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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