

Sleep Apnea

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In order to understand what sleep apnea is, it is important to know what normal sleep is. Normal, uninterrupted sleep consists of a cyclic pattern alternating between rapid eye movement (REM) sleep and non-rapid eye movement (non-REM) sleep. REM sleep is also called dream sleep. During REM sleep there are many physiological changes observed. For instance, there is a decline in chin muscle activity, generalized muscle atony (relaxation) except the diaphragm, and irregular breathing. Consequently, with the onset of normal sleep, the pharynx (airway) narrows because of muscle relaxation, causing added resistance to air movement through the airway. During normal sleep, this causes reduced air movement and a slight increase in carbon dioxide in our body.

What is sleep apnea? Sleep apnea is defined as a complete cessation of breathing from any cause during sleep, resulting in decreased oxygen in the blood or increased carbon dioxide (a greater increase than would be seen in normal sleep). The pauses in breathing usually last 10 to 20 seconds but can last as long as 120 seconds. In severe cases, more than 500 attacks of sleep apnea may occur during a night.

Sleep apnea is divided into 2 major types. The most common is obstructive sleep apnea, which is caused by obstruction of the airway. This is the type that is believed to be more common in people with Down syndrome. Respiratory effort continues but the obstruction prevents movement of air into and out of the lungs. The less common type is non-obstructive (central) sleep apnea. In this type, there is a problem in the brain or nervous system that controls our breathing. The airway may be open and the muscles may be fine but the signal to breathe is not sent or doesn't reach the muscles.

Sleep apnea is increasingly recognized as a significant cause of health problems and even death. More than one percent of the general population is affected, with a dramatic increase in the elderly. Approximately 4% of middle-aged men have obstructive sleep apnea whereas women are much less frequently affected. Obesity is the major risk factor although it is not invariable (many obese people do not have sleep apnea and some people with sleep apnea are not obese). Typically, conditions that narrow the upper airway, such as enlargement of the soft palate, uvula, tonsils, adenoids, local fat deposition, enlarged tongue and other structural abnormalities predispose to sleep apnea.

Additionally, persons with Down syndrome are predisposed to obstructive sleep apnea because their relatively small midfacial region with a relatively large tongue contributes to the compromised airway. Narrowing of the opening of the skull for the nasal passages and narrowing in the airway below the pharynx may also contribute to a diminished airway. Chronic inflammation of the nasal passages, enlarged lymph tissue (including the tonsils and adenoids) and obesity, which are common in persons with Down syndrome, are added risk factors. Maintenance of an open airway is dependent on the muscle tone in the pharynx. Muscle tone in this area is often reduced in persons with Down syndrome as part of a generalized decline in muscle tone. The use of sedative medications and, perhaps, antihistamines may also contribute to decreased muscle tone.

Characteristically, a long history of loud snoring combined with restless sleep, excessive daytime drowsiness, and early morning headaches suggest the diagnosis of sleep apnea. Also there may be an inability to concentrate, depression, irritability and personality changes. During obstruction, there may be aspiration of secretions into the lungs causing a cough or aggravating asthma symptoms. As the problem progresses, shortness of breath and fatigue increase.

The diagnosis of apnea begins with recognition of the presenting symptoms. Hypothyroidism (underactive thyroid) should be excluded. The physical exam usually, but not always, reveals obesity and excessive soft tissue in the mouth, pharynx and neck. With advanced disease, the right side of the heart is weakened and findings of failure of that part of the heart may be seen. The laboratory tests are usually normal except low oxygen and high carbon dioxide may be found.

An overnight sleep study is needed to definitely diagnose sleep apnea. It involves recording eye movements, muscle tone, electroencephalogram (EEG) to measure brain waves, and electrocardiogram (EKG) to measure the electrical activity of the heart. The test also records respiratory movements, nasal and oral air movement, and oxygenation of the blood.

If obstructive sleep apnea is found, there are several alternatives for treatment. If the patient is overweight, the first consideration is to lose weight. Many people have less or no sleep apnea if they don't sleep on their back. Therefore, encouraging the person to sleep on their side or on their abdomen can be helpful. Sometimes placing a sock on the back of the pajama top and putting a tennis ball in the sock keeps a person off their back. Eliminating sedatives and alcohol also helps some people.

Oxygen by a nasal cannula (the two-pronged device commonly used in the hospital to deliver oxygen) can help some individuals. However, generally if the oxygen is to get to the lungs, oxygen alone is not enough. For those individuals, CPAP (continuous positive airway pressure) is used. This is delivered by a mask that fits over the nose or the mouth and nose. It delivers a positive pressure to the airway to keep it open.

For some individuals, surgery to open the airway is indicated. Removal of nasal polyps, correction of a deviated septum or removal of enlarged tonsils and adenoids can sometimes help. However, often there is also obstruction in other parts of the upper airway and a more extensive surgery is required.

Uvulopalatopharyngoplasty is a surgery that removes the uvula, redundant soft palate tissue, tonsils, and adenoids. When there is no other available treatment, some patients require a tracheostomy (hole in the neck) to allow breathing around the obstructed airway.

If sleep apnea is not treated, serious complications can arise. In addition, the chronic sleep deprivation of sleep apnea and the poor oxygenation can lead to significant mood and behavioral changes that can be misinterpreted as purely psychological. Care must be taken in these instances because a medication with sedative effects may be prescribed to help with these changes. The sedation could make the sleep apnea worse which could cause an increase in mood and behavior disturbance.

People with Down syndrome have a higher incidence of sleep apnea. Awareness of the possibility of sleep apnea and observation for the symptoms of daytime drowsiness, disturbed sleep, and pauses in breathing while sleeping are the first steps towards making the diagnosis.