Obstructive sleep apnea in children with Down syndrome

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What is obstructive sleep apnea (OSA)?

Sleep apnea is a pause in breathing while sleeping. In obstructive sleep apnea there is obstruction, or blocking of the upper airways (the tubes between the mouth and the lungs), preventing air from entering the lungs. This is different from central sleep apnea, in which there is a pause in breathing because the brain does not send a signal to the body to breathe.

What happens during normal breathing?

A signal is sent from the brain to the respiratory (breathing) muscles. The largest of these is the diaphragm, found at the base of the chest. When the diaphragm contracts, it pulls downward toward the abdomen, causing "negative pressure" that sucks air in through the airways and into the lungs. In the lungs, oxygen is absorbed from the air into the bloodstream, and carbon dioxide is released from the bloodstream into the air to be exhaled. At the end of the breath the diaphragm relaxes, allowing the air to be exhaled.

What happens in Obstructive Sleep Apnea?

The negative pressure inside the chest while breathing in has almost no effect on the windpipe and voice box, which are stiff and hardly change in dimension. However, above the voice box, the upper airway is a tube made of soft tissue, including skin, tonsils, adenoids, tongue, muscle, and fat, which gets sucked inwards by the negative pressure. If the upper airway is crowded, it becomes too narrow, restricting airflow and resulting in partial or full collapse of the upper airway. To visualize this, try sucking on a straw with your mouth over one end and your finger partially covering the other end. As you slowly cover the end with your finger you will notice it becomes more difficult to suck in the air. At a certain point, the negative pressure within the straw will overcomes the straw's ability to remain open, and it collapses inward, limiting or fully stopping all airflow through it.

In obstructive sleep apnea, the collapse of the upper airway prevents air from adequately entering into the lungs until the obstruction is relieved. This can cause changes in both the blood oxygen (which drops as too little oxygen gets into the lungs) and carbon dioxide levels (which rise as too little carbon dioxide is removed from the lungs). The episodes of obstruction usually end with a physical arousal, a disruption in the sleep patterns, or both, which result in fragmented and interrupted sleep.

Why doesn't the upper airway obstruction occur during wakefulness?

In some children, partial upper airway obstruction does occur while they are awake. These children often sound congested, and breathe better in certain body positions. However, once asleep, muscle tone is more relaxed than while awake, particularly in the stage of sleep called REM (rapid eye movement sleep). This relaxation of muscle tone causes the size of the upper airway to become smaller, which leads to worsening collapse while breathing in and to either partial or complete obstruction.

What does this have to do with Down syndrome?

OSA is quite common in the general pediatric population, affecting between 2-5% of children. It is much more common in children with Down syndrome, with studies showing OSA in between 30-60% of children with Down syndrome. There are many reasons why children with Down syndrome are more susceptible to obstructive sleep apnea. The upper airways are usually smaller (in proportion to their size) than those of children without Down syndrome, because of a larger tongue and smaller maxilla (upper jaw). Other factors can contribute to a narrower airway passages, such as lower muscle tone, irritation of the soft tissues of the upper airway because of GE reflux, and different patterns of fat deposition in the neck.

OK, so my child snores. Why is this a problem?

OSA is a serious health problem at any age. In adults, OSA is known to cause high blood pressure, poor sugar tolerance, increased incidences of heart disease and stroke, pulmonary hypertension, and right-sided heart failure. In children, in addition to all of these, it can also cause poor growth, delayed development, decreased school performance, and increased attention problems. These are important concerns in any child, and can be even more significant in a child with an underlying developmental delay.

How do I know if my child has OSA?

Most children with OSA snore, and this snoring is often accompanied by pauses in breathing, snorting, choking and gasping for breath. Children with OSA often breathe with their mouths open, and sleep with their necks extended in an effort to open their upper airways and allow for better passage of air through them. Sometimes a noticeable change in nighttime breathing patterns is associated with changes in behavior or in school performance. Children who have OSA can complain of waking up with a dry mouth or headaches in the morning. In some children, nighttime bed wetting returns after months or years of the child being dry at night.

The only way to be sure whether or not a child has OSA is by doing a sleep study. This involves spending a night in the sleep lab (accompanied a parent), during which the child sleeps hooked up to monitoring equipment that measures different physiologic data. After analyzing these data, a sleep specialist can determine whether or not the child has OSA, and if so, how severe it is.

If my child has OSA, what happens?

Most children with OSA are evaluated by an Ear, Nose & Throat specialist for removal of the tonsils and the adenoids. While this surgery is generally curative in about 85% of children, the success rate in children with Down syndrome is often not as high. If your child does undergo surgery, she or he may need to have a follow up sleep study 6-8 weeks afterwards, to make sure that the surgery has brought about full resolution of the upper airway obstruction.

If there is still obstruction after surgery, or if your child is not a candidate for removal of the adenoids and tonsils, he or she may be referred back to the sleep lab for a continuous positive airway pressure (CPAP) titration study, or for other surgical interventions, if warranted.

What is CPAP?

CPAP stands for continuous positive airway pressure. It is given by a machine which blows air into the upper airways at a pre-set, constant pressure. The machine is attached by an air hose to a mask worn on the face while sleeping. The positive pressure expands the upper airway and prevents its collapse. The optimal pressure setting is identified during a titration (adjustment) study, during which different pressures are trialed to determine the lowest pressure needed to successfully prevent obstruction. While CPAP can seem cumbersome at first, most children, including those with special needs, successfully adapt to it, and learn to associate wearing it with feeling better during the day. Close follow up is important to make sure that the CPAP is being used correctly and that difficulties using it are addressed quickly in order to prevent its use from becoming unpleasant to the child.

If my child is prescribed CPAP, does that mean that s/he will need it forever?

No. As OSA is a disease process resulting from the interaction of many factors, it is possible that with time some of these may lessen. For example, OSA in a child who is overweight may lessen or completely resolve with weight loss. On the other hand, the degree of OSA present can also worsen with time, requiring the use of CPAP at higher pressure settings. This is why children who are on CPAP need to be seen by their physician on a regular basis to ensure that they continue to receive appropriate therapy.