

Respiratory Problems in Prader-Willi Syndrome

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Several recent articles continue to show that individuals with Prader-Willi syndrome are at risk for respiratory problems. In particular problems of central hypoventilation/apnea, and obstructive apnea in Prader-Willi syndrome have recently been investigated.

Central hypoventilation is a disorder of decreased breathing rate or depth particularly during sleep. This usually causes problems with daytime sleepiness and if significant can cause problems with elevated blood pressure in the lungs. Individuals with Prader-Willi syndrome may be at increased risk for this due to decreased muscle tone and mass, excessive obesity, and possibly decreased neural drive for breathing. Studies have shown some individuals with Prader-Willi syndrome have decreased depth and rate of breathing.

Central apnea means the complete cessation of breathing during sleep. There are several studies that show an alteration in the response of some individuals with Prader-Willi syndrome to chemicals that would normally increase breathing. Both receptors in the body and the area of the brain that is involved with breathing are being investigated. The clinical significance of central apnea is still under investigation.

Obstructive sleep apnea is well known to occur in Prader-Willi syndrome as well as in other syndromes with hypotonia (poor muscle tone) such as Down syndrome. It is seen in 2% of the normal pediatric population as well. This results when the individual is trying to breathe while asleep, but due to obstruction in the airway, no air enters the lungs. The obstruction can occur anywhere from the nose to the small airway passages in the lungs. These individuals usually have loud breathing and snoring associated with periods of quiet where no air movement is noted. Untreated obstructive apnea can have serious complications including death.

Other problems that can cause respiratory difficulties in the young can be chronic stomach reflux and aspiration. Although the lack of vomiting is felt to be prominent in Prader-Willi syndrome, reflux has

been documented and should be investigated in young children with chronic respiratory problems. Individuals with obstructive apnea are at more risk for reflux as well.

The American Academy of Pediatrics has recently set forth guidelines for diagnosis and management of obstructive sleep apnea. The guidelines suggest that all children be screened with history of snoring or other evidence of airway obstruction. Your physician may wish to obtain a sleep study if there is excessive sleepiness, significant obesity or before surgery. In those individuals with a positive history, a sleep study is performed where breathing patterns, heart rate, oxygen levels and air movements are recorded. If the test is positive, further evaluation may need to be performed to individualize the treatment. The primary treatment as suggested by the guidelines would include tonsillectomy and/or adenoidectomy or CPAP (Continuous Positive Airway Pressure), where the individual wears a mask at night to keep the airway open.

Frequently obstructive and central apnea may occur in the same patient. This is probably true in the majority of individuals with Prader-Willi syndrome with respiratory problems. Both obstructive and central apnea can be evaluated by a sleep study.

In summary, individuals with Prader-Willi syndrome are at risk for respiratory problems, most commonly obstructive apnea. If any child has symptoms of obstructive apnea, a sleep study should be obtained. The role of central apnea in Prader-Willi syndrome is under investigation.

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Diagnosing and Treating Sleep Apnea in Children and Adults with Prader-Willi Syndrome

By Barb Dorn, RN, BSN

More information and knowledge is being learned about apnea and Prader-Willi syndrome (PWS). Apnea is a condition in which a person stops breathing for a prolonged period of time while sleeping. Over time, this can result in high blood pressure, heart attack and/or stroke. There are two types of apnea – central apnea and obstructive apnea. The problem in central apnea arises in the brain. The cause for this is often unclear; however, the message to breathe is altered, resulting in the stopping or interruption of breathing. In obstructive apnea, a person's throat or airway passage becomes blocked; air can't flow to the lungs. Many persons with PWS have been found to experience obstructive apnea. Risk factors for obstructive apnea include obesity and poor muscle tone. The symptoms of obstructive apnea include snoring, daytime sleepiness, waking up feeling tired and unrested, morning headaches, difficulty concentrating and, irritability and/or agitation. 90% of persons with PWS experience daytime sleepiness. Previously, obesity was believed to be the primary reason for this breathing problem. However, this problem is being documented in persons with PWS who are not obese.

Researchers have found that persons with PWS show other breathing and sleep abnormalities. Sleep studies have shown delayed sleep onset, frequent arousal in sleep, an increase in total waking after sleep onset and Rapid Eye Movement (REM) abnormalities (Vela-Bueno, Kales & Soldatos, 1984; Harris & Allen, 1985; Hertz & Cataletto, 1992). Other breathing problems in persons with PWS have been noted, including mild obstructive apnea, lowered oxygen levels during sleep, and abnormal responses when oxygen and carbon dioxide levels were altered. When a person breathes too shallow or if their throat is obstructed, their oxygen level falls and the carbon dioxide level rises. Normally, our body responds by either increasing the rate of breathing or by causing a person to move or arouse (which then makes them breathe deeper). It has been found that persons with PWS do not increase their breathing or arouse when these levels are abnormal during sleep (Mendez, 1990).

Sleep apnea is diagnosed with a sleep study. Various sensors and electrodes are attached to monitor airflow, heart rate, leg movements, oxygen levels as well as brain waves. Sleep studies are done at night and can be done in a hospital or at home. The application of the sensors and the need for cooperation for a prolonged period of time can often be challenging for the person with PWS. Because of their tenuous emotional control, this process may lead to anxiety and/or emotional escalation. Some persons with PWS are also sensory defensive and become very anxious when devices are attached to them. In order to decrease family stress and to maximize cooperation, in-patient testing is often recommended. Persons with PWS are often "people pleasers" and cooperate more with technicians than they do with family members. Admission to the sleep laboratory is typically late in the evening with discharge in the early morning.

Maximizing Success with a Sleep Study Behavior Management Strategies

- **Foreshadow what to expect.**
- **Sleep technician may need to allow extra time for breaks. Apply electrodes – take a break – have a stretch – verbal praise – apply more.**
- **Use diversion while applying sensors. Watching a video may be helpful.**
- **Provide lots of praise!**
- **Celebrate success; reward when completed. If possible, take a photo after all electrodes are applied. It is a big hit for "show and tell".**
- **For those who are sensory defensive, have person wear some electrodes a day or two before test. It may decrease sensitivity and anxiety.**
- **Inform physician of problem. Priorities may need to set in what is to be assessed.**
- **Apply most sensitive area last.**

If the person with PWS is found to have sleep apnea, measures to correct this must be started. Obesity is the number one correlate to sleep-related lowered oxygen levels. Weight management must be instituted. A nutrition referral and/or evaluation of environmental supports may be needed. Exercise should also be a part of this program.

To treat obstructive sleep apnea, the physician may recommend an air pressure device called CPAP (Continuous Positive Airway Pressure). This treatment device delivers gentle, steady airflow through a soft mask that is placed over the nose. This helps to keep the throat structures from blocking the airway. The physician will determine the amount of pressure that is needed. It is often difficult to adjust to wearing this mask while sleeping. The most successful approach is to be helpful and encouraging – not adversarial.

Surgery may also be suggested to remove excess tissue or enlarged tissue that may be causing obstruction of the throat.

In summary, persons with PWS are at increased risk of developing obstructive sleep apnea. Because of behavior challenges, diagnosing and treating this disorder can be problematic for these individuals, as well as for those who support them. Treating sleep-related breathing problems can help the person with PWS awake, feeling alert and refreshed. It may help prevent daytime sleepiness and other serious health problems.

Tips for Using CPAP for the Person with PWS

- **Work on increasing the wearing time of the mask, gradually. Start with wearing the mask 5-10 minutes before bedtime. Slowly increase time – 10 minutes the first few nights, 15 minutes the second night. It may take a month before wearing it all night.**
- **Celebrate success. Use lots of verbal praise. Use an incentive chart if needed.**
- **If needed, give the person with PWS some control. Allow one night off a week – his/her choice.**
- **If person refuses to wear, do not argue or force. Point out positive outcomes – less headaches, more rested ...**
- **Have weekly “check-ins” with the nurse. The nurse can offer encouragement to person with PWS.**