

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 movement 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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