



Prader-Willi Family NEWSLETTER

Spring 2022 Edition

Endocrine Corner: Considerations for Excessive Sleepiness and Fatigue in PWS

by Sarah Hu, CPNP

Prader Willi Syndrome (PWS) is a complex genetic condition that affects many systems of the body and leads to distinct characteristics/features. Many of these issues are thought to be related to dysregulation of the hypothalamus. The hypothalamus is a control center in the brain that helps to regulate hormone production, temperature regulation, sleep, pain sensitivity and hunger/fullness queues.

Easy fatigue, low endurance and excessive daytime sleepiness are frequently reported symptoms among Prader Willi patients. We will discuss below conditions that our Prader Willi patients are at risk for, surveillance and treatment.

Hormone deficiencies

- **Thyroid Hormone:** thyroid hormone is an important hormone that has many functions throughout the body. The general function is regulating energy expenditure. Low thyroid hormone levels (hypothyroidism) can be associated with increase fatigue, constipation, and weight gain. We generally recommend screening thyroid function 1-2x a year.
- **Cortisol Hormone:** Cortisol is an important hormone made by the adrenal gland that helps the body during times of stress by activating many compensating processes in the body. Cortisol deficiencies can lead to increase fatigue, poor growth, weight loss, weakness, and poor recovery from illness. Due to hypothalamic dysregulation-children with PWS may be at risk for central adrenal insufficiency which leads to low cortisol secretion due to inadequate signals from the brain to the adrenal gland. We generally recommend screening morning cortisol levels every 1-2 years or if symptomatic.

Sleep disorders/Sleep Apneas: conditions in which breathing periodically stops and starts during sleep. Untreated sleep apneas can lead to excessive daytime sleepiness, poor cognitive function and focus, hypertension, and worsening obesity.

- **Central Sleep Apnea (CSA):** This type of sleep apnea is called “central” because the brain is not sending the proper signals to the muscles that control your breathing. Infants with PWS have a higher frequency of CSA which is why sleep/nap studies are recommended early on to screen.
- **Obstructive Sleep Apnea (OSA):** This type of sleep apnea more common than CSA. It is called “obstructive” as the breathing/ventilation pathway is blocked. Children and adults with PWS are at high risk for OSA due to enlargement of tonsils and adenoids (which can be worsened by growth hormone), low muscle tone that helps with opening the airway passage, and obesity.

Sleep studies are recommended before and after growth hormone start, prior to spinal fusion surgery or if symptoms/increasing risk factors are present. There are still reports of daytime sleepiness/hypersomnia, easy fatigue, and low endurance in those who do not have the above hormone deficiencies or sleep disorders (or are being appropriately treated). The exact cause(s) is still not well understood and is attributed again to dysregulation of the hypothalamus.

A very small clinical trial of 9 PWS subjects published promising results in 2011 for use of Modafinil for excessive daytime sleepiness. However, a larger and more rigorous clinical trial for this medication has not been pursued and thus this medication is not routinely used. There is a current and ongoing clinical trial (Phase 2) for Pitolisant as treatment of excessive daytime sleepiness in PWS subjects.



Behaviorist's Corner: Melatonin

By Dr. Peter Chung

Melatonin is a hormone that is naturally produced by the brain to regulate the day-night cycle (also called the circadian rhythm). If you've ever traveled to a different time zone and had difficulty with your body adjusting, it's often due to disruption of your body's natural day-night rhythm. Natural melatonin production is affected by a variety of factors, including daily activities like eating and exercise. Light from external sources, especially blue light that comes from electronic devices, can "trick" the brain by mimicking actual sunlight. There are some medical diagnoses and genetic syndromes (for example, Smith-Magenis Syndrome) in which melatonin production can be affected, resulting in sleep disturbances.

Melatonin is available as an over-the-counter supplement. As such, it is not regulated by the Food and Drug Administration. This means that different versions of melatonin may have different strengths, concentrations, and purity levels. Because melatonin is a supplement, formal research studies on its use may not generalize to everyday usage (e.g. the melatonin used in a research study may not be the exact kind of melatonin you find in your pharmacy). People will sometimes use melatonin to help for the short-term treatment of sleep-onset insomnia (difficulty falling asleep). Research has shown that supplementation can be helpful for some children with sleeping problems, including neurodevelopmental conditions like autism spectrum disorder and attention-deficit/hyperactivity disorder (<https://pubmed.ncbi.nlm.nih.gov/29720494/>). However, it is important to recognize that many of these studies were small and limited to a short time period. It is best to discuss any potential use of melatonin supplements with your health care provider.

There is limited research on Prader Willi syndrome and melatonin production. One study (<https://onlinelibrary.wiley.com/doi/10.1002/ajmg.a.33001>) found that individuals with PWS did not differ from the general population on morning melatonin levels, although anti-depressant medications were linked to higher morning melatonin levels. The Journal of Clinical Sleep Medicine has recommended that additional research be conducted on sleep, sleep disorders, and sleep treatments for individuals with PWS. <https://jcs.m.aasm.org/doi/10.5664/jcs.m.9938>

Recipe Corner:

Healthy Banana Ice Cream



Ingredients:

- 1 Small Banana
- 1 Tbs Unsweetened Vanilla Almond Milk

Directions:

1. Cut ripe banana into 4 pieces and freeze
2. Add frozen bananas and unsweetened vanilla almond milk to food processor or blender
3. Blend until smooth and portion into 3 containers.

1/3 of recipe: 30cal, 6.7g carb, 0g fat, 0.3g pro.

Optional: Swap out the banana for your favorite fruit, such as strawberries or blueberries, to create a variety of flavors!

We have exciting news from our friends from the Pulmonary Department!

SLEEP CENTER

Our two newly expanded, state-of-the-art sleep centers offer:

- An 8-bed, 3,200-square-foot facility conveniently located across the street from CHOC Hospital in Orange
- A 4-bed facility inside CHOC at Mission Hospital in Mission Viejo
- Accreditation by the American Academy of Sleep Medicine
- Ranking as one of the top pulmonology and neurology programs in the nation by *U.S. News & World Report*
- Equipment specifically designed for pediatric sleep studies
- Serene, child-friendly environments depicting calming nature scenes
- Private bedrooms in the Orange location, each equipped with an additional bed to allow a parent or guardian to stay during the study
- A multidisciplinary team of board-certified sleep medicine pulmonologists and neurologists, psychologists, respiratory therapists and sleep technologists, all specially trained to work with infants, children and adolescents
- Close collaboration with CHOC clinics and programs, including otolaryngology, plastic surgery and more
- Comprehensive sleep study capabilities to evaluate breathing, brain activity, heart activity, blood oxygen and carbon dioxide content, chest and abdominal wall movement, muscle activity and the amount of air flowing through the nose and the mouth
- CPAP/BiPAP titration studies to fit for a proper mask size and calibrate air flow while the child sleeps
- Outpatient specialty clinic care for kids who require evaluation and treatment for all sleep related issues.

CHOC Sleep Center

530 S. Main St., Ste. 100, Orange, CA 92868

CHOC at Mission Hospital Sleep Center

27700 Medical Center Rd., Mission Viejo, CA 92691

Learn more about the CHOC Sleep Center at choc.org/sleepcenter.



Social Worker's Corner: Tidbits for a good night's sleep

By: Bobbi McGann, LCSW

The following recommendations will help your child get the best sleep possible and make it easier for them to fall asleep and stay asleep:

- **Sleep schedule.** Your child's bedtime and wake-up time should be about the same time every day. There should not be more than an hour's difference in bedtime and wake-up time between school nights and non-school nights.
- **Bedtime routine.** Your child should have a 20 to 30-minute bedtime routine that is the same every night. The routine should include calm activities, such as reading a book or talking about the day, with the last part occurring in the room where your child sleeps.
- **Bedroom.** Your child's bedroom should be comfortable, quiet, and dark. A nightlight is fine, as a completely dark room can be scary for some children. Your child will sleep better in a room that is cool (less than 75° F). Also, avoid using your child's bedroom for time out or other punishment. You want your child to think of the bedroom as a good place, not a bad one.
- **Caffeine.** Your child should avoid caffeine for at least 6 to 8 hours before bedtime. Caffeine can be found in many types of soda, coffee, iced tea, and chocolate.
- **Evening activities.** The hour before bed should be a quiet time. Your child should not get involved in high-energy activities, such as rough play or playing outside, or stimulating activities, such as computer games.
- **Television.** Keep the television set out of your child's bedroom. Children can easily develop the bad habit of "needing" the television to fall asleep. It is also much more difficult to control your child's television viewing if the set is in the bedroom.
- **Computers, IPADS and phones.** Children should not be on any devices prior to bedtime. If they are using these devices in the evening, make sure you go under settings to change device to nightshift several hours before bedtime.
- **Naps.** Naps should be geared to your child's age and developmental needs. However, very long naps or too many naps should be avoided, as too much daytime sleep can result in your child sleeping less at night.
- **Exercise.** Your child should spend time outside every day and get daily exercise, but, if possible, limit exercise within 4 hours of bedtime.
- **Parents should follow many of the items listed above for better sleep as well!**





Dietitian's Corner: How Your Child's Nutrition Impacts Their Sleep Pattern

By Christina Wright-Yee, MPH, RD, CSP & Maria Solfrizzo, Cal Poly Pomona Intern

Sleep is essential for growing children. Having a routine sleep schedule is especially important for children with Prader-Willi syndrome as it can positively influence nutrition. A lack of good, quality sleep can:

- Slow metabolism and increase the risk of obesity.
- Increase cravings for carbohydrates (carbs) and other high calorie foods, such as French fries, chips, pastries, candy, and sugar sweetened drinks.
 - Consistent high-carbohydrate meals have been shown to impair sleep quality by increasing the number of awakenings at night and reduce the amount of deep sleep.
- Affect the way our body breaks down food into sugar to use for energy, increasing insulin resistance. This can lead to the development of type 2 diabetes.
- Be possibly related to your child's lack of intake of potassium, calcium, magnesium, and vitamins A, C, D, E, and K. These nutrients are often found in fruits, vegetables, and healthy fats. Be sure to discuss these with your dietitian.

You and your family can improve quality of sleep by:

- Creating a normal sleep schedule provides structure which can help establish a mealtime routine throughout the day and limit late night eating. It also reduces opportunities for snacking to avoid consuming extra calories.
- Increasing fruits and vegetables with meals or snacks, while limiting salt and saturated fats.
- Doing daily exercise to use up any energy. Plan the last physical activity for the day about 90-120 minutes before bed to give their body plenty of time to cool down before going to sleep.
- Limiting caffeine and sugary foods. Avoid caffeine for at least 6-8 hours before going to bed. Soda, iced tea, coffee, and chocolate may contain caffeine.

Follow these tips to create and maintain a routine sleep schedule at night. This will help to promote healthy nutrition during the day.

Please note that May is Prader-Willi Awareness Month!

Sleep & Nutrition

Prader-Willi Syndrome

Sleep is so important for growing kids! It promotes healthy eating habits, growth, and improves metabolism.

A lack of sleep affects hunger and can cause children to overeat at meals and snack throughout the day. We begin to crave carbohydrates, and children will seek out high calorie carbs such as french fries, chips, desserts, and soda.

Poor sleep changes the way our bodies break down these carbs into sugar, which can lead to type 2 diabetes.

**Avoid caffeine 6-8
hours before bedtime**

Create routine bedtime

**Exercise 2x/day
Last activity 90-120
minutes before bed**



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