WHAT IS PRADER WILLI SYNDROME?

Prader-Willi Syndrome (PWS) is a complex genetic condition that can affect 1 in 15,000 people. In infancy, it is characterized by weak muscle tone, poor feeding, and delayed growth and development. As children get older, it can be associated with behavioral issues, short stature, hormonal abnormalities, and chronic overeating that can result in obesity. It is also associated with different sleep and breathing challenges that should be understood and managed to optimize health and quality of life.

WHAT ARE SLEEP-RELATED BREATHING DISORDERS?

Obstructive sleep apnea (OSA) is a condition in which children have frequent pauses in breathing during sleep (Figure 1). Symptoms of OSA can include snoring, pauses in breathing while asleep, frequent awakenings from sleep, restless sleep, and daytime sleepiness. Children with OSA can have some or all of these symptoms. OSA is diagnosed by performing an overnight sleep study (polysomnogram) in a sleep laboratory. OSA can lead to poor sleep quality and health problems that can affect your child’s ability to learn, grow, and develop. OSA can cause low oxygen levels and, over time, can cause strain on the heart.

If there are frequent pauses in breathing during sleep that are not due to airway obstruction, the cause may be central sleep apnea. This is a diagnosis made by sleep study. Infants with PWS are at increased risk for central sleep apnea, which can either resolve or persist as children get older. Some patients who have central sleep apnea during infancy can develop OSA later (1).

Other breathing problems can occur during sleep, including low oxygen levels (called hypoxemia) or high carbon dioxide levels (called hypoventilation) (2). Hypoventilation can contribute to headaches, foggy mind, and fatigue. Children with PWS may have abnormal responses to these breathing problems and may not able to adapt their breathing pattern when needed (3).

WHY DO CHILDREN WITH PWS HAVE INCREASED RISK FOR OSA?

It is estimated that up to 80% of children with PWS have OSA (4), in contrast to 2–3% of all children. This increased risk is due to a combination of factors, including differences in facial and upper airway anatomy, decreased muscle tone (causing airway collapse when sleeping), airway obstruction by the tongue, and obesity.

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There are several potential benefits of growth hormone (GH) therapy in children with PWS, including improved muscle tone and development. Historically, there has been concern that GH therapy might increase the risk of OSA in children with PWS, with some guidelines recommending that patients should not receive GH therapy if they have severe OSA that is not treated (5). More recent studies show that GH treatment does not cause OSA in most patients, so many providers feel that GH treatment can be started early and safely with close monitoring (6). Your child’s provider may consider performing a sleep study a few months after starting GH to reevaluate for OSA. A sleep study is also recommended if new symptoms of OSA are seen.

WHAT IS DRUG-INDUCED SLEEP ENDOSCOPY AND WHEN IS IT PERFORMED?

When the cause for OSA is not completely clear and/or positive airway pressure (PAP) is not tolerated or effective, your provider may recommend a drug-induced sleep endoscopy (DISE). During DISE, the child is placed under anesthesia to simulate sleep, and while asleep, the ear, nose, and throat surgeon can look in the airway with a camera for sites that can be addressed with surgery. Studies using DISE in children with PWS have revealed that collapse of the tongue base can often cause obstruction of the airway (7).
HOW CAN YOU TREAT SLEEP-RELATED BREATHING DISORDERS IN CHILDREN WITH PWS?

There are several possible treatment options for OSA in children with PWS, including medical therapy with allergy medication, tonsillectomy and adenoidectomy (T&A) or other airway surgeries, PAP, working on fitness and nutrition to avoid obesity, positional therapy if the OSA occurs only when the child is on their back, orthodontic treatment, or supplemental oxygen in carefully selected cases. Choosing which treatment is right for your child will depend on OSA severity, the major underlying reason for airway obstruction, your treatment preferences, and the recommendations of your treating provider.

Central sleep apnea in infants with PWS can be treated with supplemental oxygen, using a nasal cannula (8). Oxygen should be started with care and your provider will typically perform a sleep study before prescribing oxygen to be used at home. Your provider may consider other treatment options, including different types of PAP therapy (including a Bilevel PAP with a backup rate, which provides advanced support) or medications that increase breathing rate (such as acetazolamide) (5, 6).

HOW WELL DOES T&A WORK FOR OSA IN CHILDREN WITH PWS?

T&A in patients with PWS and OSA helps to decrease severity of disease, with about 20% achieving total cure and 60–70% having substantial decrease in severity of OSA (9). Although surgery can decrease OSA severity, about 80% of patients with PWS continue to have some degree of OSA after surgery (1–3, 7, 8).

CAN CHILDREN WITH PWS DEVELOP NARCOLEPSY?

Many children with PWS have excessive daytime sleepiness (hypersomnia). Children with PWS are at an increased risk of narcolepsy and hypersomnia, possibly related to abnormal function of the part of the brain (the hypothalamus) that controls the body’s hormonal, metabolic, and neurologic functions (hypothalamic dysfunction) (10). In addition to daytime sleepiness, symptoms of narcolepsy include cataplexy (brief episodes of loss of muscle control associated with emotion), sleep paralysis (not able to move your body upon awakening), and sleep-related hallucinations (seeing or hearing things that are not present during sleep onset or upon awakening). Narcolepsy can be diagnosed with a daytime sleep study (called a Multiple Sleep Latency Test). Although there is currently no cure for narcolepsy, there are several medications that can help lessen symptoms and improve quality of life.

ACTION STEPS

- Talk to your child’s healthcare provider if you notice symptoms of sleep apnea (snoring, pauses in breathing, daytime sleepiness, or unrefreshing sleep).
- If your child has been diagnosed with OSA, talk to your healthcare provider about treatment options, such as surgery or PAP therapy.
- If your child with PWS is going to start GH, ask your provider about when your child should have a sleep study.
- Tell your healthcare provider if you notice symptoms of sudden loss of muscle control, sleep paralysis, or hallucinations, as these may be signs of narcolepsy.

Author disclosures are available with the text of this article at www.atsjournals.org.
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