

PWSA | USA Presents:

Medical Reference Guide for Parents




Prader-Willi
SYNDROME ASSOCIATION | USA
SAVING AND TRANSFORMING LIVES

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PWSA | USA gratefully acknowledges the printing of this booklet is funded by Lota and David Mitchell in loving memory of their son, David Scott Mitchell, brother of Julie Mitchell, who has PWS.

What is Prader-Willi Syndrome?

Prader-Willi syndrome (PWS), pronounced PRAH-der-WILL-ee, is a birth defect that results from inactive or missing genes on chromosome 15.

PWS occurs in approximately one in 12,000-15,000 births and equally affects both sexes and all races.

The major characteristics of PWS include low muscle tone, poor growth, learning difficulties, incomplete puberty, an excessive drive to eat, metabolic disturbances, and difficult behaviors.

The major medical concern of PWS is morbid obesity.

For each of the Developmental and Medical Concerns listed in this brochure, PWSA | USA has resources to help parents and care providers manage, reduce, and in some cases even eliminate the symptoms of PWS.

Cause and Diagnosis of PWS

Prader-Willi syndrome is a genetic disorder but is generally not inherited. There is no known cause of PWS and there is nothing that either parent did or didn't do to cause their child to have the syndrome.

PWS results when there is a loss of genes on the paternal chromosome 15. This occurs most commonly in one of three ways: 1) Deletion of genes, which occurs in about 70% of cases; 2) Uniparental disomy (UPD) which occurs in about 25% of cases when there are two maternal chromosome 15s and no paternal chromosome 15; and 3) when there is an imprinting error, which occurs in 2-5% of cases. There is the potential for PWS to be inherited when there is an imprinting error.

All children in whom PWS is suspected should have genetic testing. If the diagnosis is confirmed, it is recommended that the family receive genetic counseling. The risk of PWS reoccurring in the same family is rare and happens only when there is an imprinting error.

With an accurate diagnosis, parents can provide appropriate and timely life-enhancing interventions.

Developmental Concerns

Growth

Most newborns are extremely lethargic and have little interest or ability to breast or bottle feed. Many infants require special bottles or nipples (e.g., Habermann feeder, NUK nipple). Growth hormone medication may improve muscle tone enough to improve feeding. A nasogastric tube (NG-tube) or a gastrostomy tube (G-tube) may be used temporarily to ensure proper nutrition. Infants should be closely monitored to ensure proper weight gain, linear growth, and head circumference growth.

PWS causes growth hormone deficiency which results in weaker muscle tone, decreased bone strength, short stature, smaller hands and feet, and increased body fat. Growth hormone therapy is now considered the standard of care for PWS and will help improve linear height (if taken prior to growth plate closure), improve muscle tone, and improve bone mineral density.

Growth hormone treatment (GHT) should be discussed when the diagnosis is made. Evidence suggests that GHT continued throughout the lifetime may have benefits on muscle tone and quality of life but testing to evaluate GH sufficiency is necessary once linear growth has been completed.

Physical Development

Developmental milestones are typically delayed by one to two years. These delays are often shortened with growth hormone therapy. Muscle tone improves as the child ages even without medical intervention. Problems with strength, coordination and balance often continue. Physical and occupational therapy should begin as soon as possible and should focus on sensory integration to improve balance and coordination. Foot and/or ankle orthotics can help support weaker ankles, especially while learning to walk. Daily physical activity and exercise are critical to weight management and overall health.

Learning and Cognitive Development

Interacting and playing with infants is critical. An Infant Stimulation therapist can provide helpful strategies. Persons with PWS typically have learning problems. Specialized testing can reveal their unique learning strengths and styles and challenges. Common strengths include long-term memory, rote memory, and receptive language skills. Common areas of concern include poor short-term memory, difficulties with abstract thinking and problem solving. Most children with PWS attend public schools and receive supports as determined in their Individualized Educational Plan through special education services. By federal law, the core therapeutic interventions – physical therapy, occupational therapy, speech and language therapy, and social skills therapy – should be available and covered by your local school system and should begin as early as possible to improve overall development.

Speech Development

Low muscle tone can interfere with babbling, articulation, and speech. Oral Motor/Speech Therapy should begin in infancy to help with feeding issues and early speech development. An assessment should be made for a diagnosis of Childhood Apraxia of Speech. Sign language and picture communication boards can aid communication and reduce frustration. Products designed to reduce dry mouth symptom may improve articulation (as well as improve overall oral hygiene).

For each of the Developmental and Medical Concerns listed in this booklet, PWSA | USA has resources to help parents and care providers manage, reduce, or even eliminate symptoms of PWS.

Social and Emotional Development

Most persons with PWS are highly social and have a strong desire for friendships. Social skills must be explicitly taught using strategies that are more concrete such as Social Stories, video, and role playing. Teaching how to understand another's perspective can be challenging but is critical. Adult intervention is often necessary to help children and adults with PWS develop and sustain meaningful relationships.

Sexual Development

Sex hormone levels (testosterone and estrogen) are typically low, causing smaller genitalia in girls and boys. Undescended testes in boys may require HCG injections and/or Orchiopexy surgery. The development of early pubic hair is common, but complete puberty is usually late and incomplete. Sex hormone therapy to advance teens through puberty remains understudied and varies from family to family.

Medical Concerns

Hyperphagia

The excessive drive to eat with a lack of satiety is the hallmark symptom of PWS that can, if unmanaged, lead to life-threatening weight gain even on a low-calorie diet. Additional information about hyperphagia is contained elsewhere in this brochure.

Stomach and Bowel Problems

Many persons with PWS have gastroparesis which is a slow emptying stomach, and a slow emptying bowel. Food is not broken down, metabolized, and eliminated from the body at the rate it should which can place excessive pressure on the stomach and/or intestinal tract, especially if too much food is consumed. Rather than localized pain, there may be stomach bloating and a general feeling of unwellness. If an individual with PWS has these symptoms, especially after a known food binge, contact a medical

professional immediately as individuals with PWS are at greater risk for severe stomach illness including perforation, which is life-threatening. A Medical Alert Booklet is available through PWSA | USA and it is recommended to always have one with you.

High Pain Tolerance

Lack of typical pain signals is common and may mask the presence of infection or injury. Someone with PWS may not complain of pain until infection is severe and may have a difficult time telling you where the pain is. Report any slight changes in condition or behavior to a medical professional for investigation into the cause.

Breathing and Sleep Problems

Individuals with PWS may be at increased risk for breathing problems due to weaker chest muscles. Anyone with significant snoring should have a medical evaluation to look for obstructive sleep apnea. An assessment for central sleep apnea may also be warranted. Sleep problems often occur. Excessive daytime sleepiness is common, with some diagnosed with narcolepsy.

Sensitivity to Medications

Individuals with PWS may have unusual reactions to standard dosages of medications. Use extreme caution when giving medications that may cause sleepiness, especially narcotics, because longer and more severe responses may occur. Water intoxication (too much water in the body) has occurred with the use of certain medications, as well as from drinking too much fluid.

Lack of Vomiting

Vomiting rarely occurs and may signal a life-threatening problem. Seek immediate medical attention especially if vomiting follows a known food binge. Due to the excessive drive to eat and the potential for eating uncooked, spoiled or otherwise unhealthy food items, lack of vomiting is of particular concern. Medications used to induce vomiting may not work and repeated doses may cause poisoning.

Body Temperature Regulation

Unexplained high or low body temperatures are common. High body temperature may occur during minor illness and in surgical procedures that utilize anesthesia. Fever may be absent even with severe illness or infection. Blood work, including a CBC (complete blood count), may be helpful to determine the severity of the illness.

Sores and Bruises

Individuals with PWS tend to bruise easily. Skin picking is common and can create open sores and the potential for infection. There are helpful strategies, techniques, and medication options to help reduce skin picking.

Other Common Concerns

- Weaker eye muscles may cause the eyes to cross (strabismus). Corrective surgery may be necessary. Talk to your optometrist about Vision Therapy prior to corrective surgery, as an option.
- Scoliosis and Kyphosis, curvatures of the spine, occur at a higher rate in persons with PWS than the general population. Curvatures can occur unusually early, can progress at a higher rate of speed, and can be difficult to detect without x-rays. Growth hormone therapy does not necessarily need to stop when treating scoliosis.
- Weaker bones (osteoporosis) may occur earlier than usual due to growth hormone deficiency and may increase risk for fractures. Growth hormone medication generally improves bone mineral density and bone strength. Bone density tests are recommended. Provide adequate amounts of calcium, vitamin D, and weight-bearing exercises.
- Dental problems are often due to dry mouth symptoms that look like thick, sticky saliva. This can lead to soft tooth enamel and significant dental and gum problems. The use of dry mouth products, such as Biotene toothpaste, increase saliva and can reduce or eliminate this symptom.

Hyperphagia

Hyperphagia, generally considered the hallmark symptom of Prader-Willi syndrome, occurs because the brain fails to regulate appetite normally. For a person with PWS there is a constant preoccupation with food accompanied by an unrelenting, overwhelming physiological drive to eat. Satiety, or the feeling of fullness when eating, does not occur as it should. Having PWS, however, does not mean individuals are destined to become obese or even overweight. With the right supports, persons with PWS absolutely can achieve and maintain a healthy body weight.

Hyperphagia generally appears subtly and in phases. From birth to about six months there is often great difficulty feeding due in part to the weaker muscle tone. As the infant ages, feeding difficulties tend to resolve through age 1 year.

Between 20 to 31 months of age there is generally increased weight gain without a significant increase in food or calories. It is during this phase (identified by Miller, et al, as "Phase 2a") that a reduction in calorie intake should often be made with the guidance of a dietician who utilizes appropriate PWSA | USA nutritional resources.

The typical PWS hyperphagia symptom usually begins around the age of 3 years as the toddler begins to show an increased interest in food. Symptoms generally increase subtly until around age 8 years when they typically level off throughout adulthood.

Beginning from Phase 2a persons with PWS require fewer calories because the metabolic rate is slower and muscle tone is typically lower. To create the proper calorie diet, consult with a registered nutritionist/dietician and use the nutrition guides available from PWSA | USA. Regular weigh-ins and a periodic diet review are recommended.

The best meal and snack plan is one that the family and caregivers are able to apply routinely and consistently. Weight control depends upon maintaining food security which requires restricting the individual's access to food. Most often, locking the refrigerator and food storage areas is the easiest method to provide the appropriate level of food security while simultaneously reducing the individual's level of anxiety related to the potential access to food.

It is important to understand that the insatiable drive to eat is a neurophysiological disorder that is not under the control of the person with PWS. There is no true ability to learn how to control hyperphagia. No medications or surgeries successfully reduce or eliminate the hyperphagia symptom – yet. There is, however, a great deal of research being done worldwide to tackle this life-limiting symptom. Until such time as there is a treatment, however, individuals with PWS must be supervised at all times and in all settings, especially where food or money to purchase food is available.

Daily Physical Activity and Exercise

Daily physical activity and exercise is essential to maintain a healthy body weight. Physical activity helps burn unwanted fat, improves muscle tone and strength, develops bone strength, keeps the heart healthy, and increases flexibility and stamina. Physical activity improves mood through the release of endorphins and decreases feelings of depression and anxiety.

Children and adults with PWS should participate in a minimum of 60 minutes a day of physical activity to maintain weight. Twice a week, activities should include strengthening exercises to build muscle and bone strength. More minutes are needed when aiming to lose body weight.

Participation in organized sports teams like Challenger Leagues and Special Olympics offers opportunities for team involvement and social play. Individualized sports activities such as swimming, karate or Tae Kwon Do offer opportunities for building strength and self-confidence. Walking for 30 minutes a day, dancing, even playing video games such as Wii Fit or Just Dance are also great ways to exercise. Whatever physical activity you choose, do it together and make it fun!

Behavior

Infants and young children with PWS are usually happy, loving, and compliant, exhibiting very few behavioral problems.

Most older children and adults with PWS have difficulty managing their behavior when feeling frustrated, disappointed, or angry. This difficulty managing negative emotions occurs because the same part of the brain that regulates appetite also regulates emotions. The shift from compliance to more tenuous emotional control usually begins when the hyperphagia symptom begins, at about age 3 years.

Anxiety is common and often underlies behavioral challenges. Reducing environmental stressors will help reduce anxiety and therefore reduce unwanted behaviors. Work to create a calm, predictable environment. Persons with PWS do best when they know what to expect and what is expected of them. Create consistent routines and schedules. Use positive rewards, incentives, and positive-toned language. Provide cues to help prepare for transitions from one activity to the next.

Most people with PWS are oppositional thinkers; the brain reflexively thinks, “No!” Provide limited, preferred choices. Don’t argue or try to talk someone out of their upset; work to let the individual know you understand their concern. Use empathy to reduce upsets.

Have patience. People with PWS typically process or think about things a bit more slowly and often need extra time to comply with a request.

Rebranded December 2022