Prader-Willi California Foundation
Supporting People with Prader-Willi Syndrome

Facts About Prader-Willi Syndrome that Conservatorship Attorneys Need to Know

Why do I need to know about Prader-Willi syndrome?

Conservatorship of the adult with Prader-Willi syndrome (PWS) is essential because without it, persons with PWS will experience potential life-threatening consequences that result directly or indirectly from the syndrome.

> **"Because of the issues PWS presents (insatiable hunger, manipulative food seeking) it is imperative that ALL food issues are NOT left up to the individual with PWS. The guardian's [conservator's] decisions on this issue alone are often the difference between life and death for the individual with PWS."**
>
> *Harry Persanis, founder and former Vice President of the Prader-Willi Alliance of NY, Inc. and father of a 39-year-old daughter with PWS*

What is Prader-Willi syndrome?

PWS is a non-inherited genetic disorder that is believed to occur because of an abnormality in the hypothalamus which affects the release of hormones from the pituitary. Symptoms occur because there are missing genes on Chromosome 15 that interfere with important regulating systems in the body including but not limited to:

- Growth
- Muscle tone
- Breathing and respiratory function
- Sleep architecture
- Temperature regulation
- Metabolism, gastric and bowel function
- Pain threshold
- Sexual development
- Cognitive function
- Emotional regulation
- Appetite regulation

Hyperphagia While all symptoms are significant, the most life-limiting and life-threatening symptom of PWS is the dysfunctional appetite regulating system which causes hyperphagia. PWS hyperphagia is a life-threatening, uncontrollable genetic drive to eat that is not satiated regardless of the quantity of food consumed.

PWS hyperphagia includes preoccupations with food; food seeking, foraging; manipulation, sneaking, hiding, and hoarding food; and eating unusual food-related items (e.g. sticks of butter, pet food, mouthwash, rotten food taken from trash). PWS hyperphagia causes food-related anxiety that frequently results in dangerous behaviors including verbal aggression, physical aggression, elopement, burglary, theft, self-injury, and lack of regard for personal safety.

There is no learning to control PWS hyperphagia. Treatment consists of restricted access to food and continuous supervision. No currently known medication reduces or eliminates this life-threatening symptom. Without 24/7 continuous support, persons with PWS will become morbidly obese or die from gastric or bowel rupture or necrosis, or from complications associated with obesity. PWS hyperphagia has caused fatality from a single food gorging incident.

Behavior Problems The second most life-limiting symptom of PWS is severe behavior problems. The PWS hyperphagia food drive underlies many food-related behavior problems but is not the sole reason for unwanted or maladaptive behaviors. Additional common symptoms include argumentative, stubborn and oppositional behaviors, impulsivity, inflexibility, obsessive and/or ritualistic behavior, excoriation (skin picking), high need for predictability and sameness, hypersensitivity to real or perceived stressors or frustrators, lying and confabulating (making up) stories, stealing (food and non-food items), and aggressive verbal and physical behaviors. The most common maladaptive behaviors include crying, yelling, screaming, shouting obscenities, hitting, punching, kicking, throwing objects, and other destructive behaviors. The need for police or psychiatric emergency team intervention is not uncommon. More than one person with PWS has been known to become so upset over a minor frustration and impulsively opened the door of a moving vehicle, jumped out, and been killed.

Why exactly do adults with PWS need to be conserved?

Irrespective of an individual's measure of intelligence (IQ), level of insight, verbal acuity, or degree of hyperphagia, persons with PWS lack adequate judgment and are not capable of making appropriate self-care decisions, *particularly* in regard to food acquisition and consumption. Adults with PWS are not capable of overruling the physiological drive to eat. Decisions made by the adult with PWS are influenced by and eventually or ultimately made in order to obtain food.

To obtain food or money with which to purchase food, adults with PWS have been known to:

- Break into neighboring homes and businesses
- Prostitute themselves or trade sex for food
- Steal valuables including jewelry to trade for food
- Run away
- Inflict harm on another
- Feign or inflict self-harm to secure hospitalization
- Make false allegations of neglect or abuse
- And many other forms of lying, cheating, and stealing in order to obtain food

Which rights are frequently conserved for adults with PWS and why?

The right to decide and fix the residence

Adults with PWS cannot make appropriate decisions regarding their residence because their decision would be driven by the syndrome's hyperphagia symptom. No one known to be diagnosed with PWS has successfully lived independently; those who have tried have either died or become morbidly obese. Whether living in the family home or outside the family home in an Adult Residential Facility setting (i.e., group home, supported living), persons with PWS require continuous, uninterrupted supervision from a staff who are specifically PWS-trained, and access to food must be restricted, most frequently with a physical lock on the refrigerator and food cupboards.

The right to access confidential records and papers

Most adults with PWS have some degree of Intellectual Disability and are unable to understand abstract and complex concepts. In addition, most people are hypersensitive to criticism or perceived criticism. Adults with PWS need a great deal of support to both access and understand or interpret records and papers.

The right to consent to marriage or a registered domestic partnership

Most adults with PWS very much want to marry and have children. Unfortunately, very few have the ability to sustain relationships. Adults often "fall in love" within hours of meeting someone, whether in person, email, text, social media. Professions of love and marriage engagements occur frequently and are broken off just as frequently. This is due in part to the syndrome's cognitive characteristics which are fairly universal amongst adults with PWS: high levels of anxiety, egocentricity, oppositionalism, impulsivity, inflexibility, need to be correct, obsessive-compulsiveness, hypersensitivity to criticism or perceived criticism, self-monitoring deficits, concrete thinking, and various executive skills deficits which typically result in upset or hurt feelings and culminate with various behavioral expressions including lying, yelling, screaming, and physical aggression. While most adults with PWS crave social relationships, most also frequently exhibit little regard for the wants, needs and interests of others when those wants, needs and interests do not coincide with their own. It is this egocentricity inherent in PWS that almost always interferes with the ability to make informed decisions about marriage. Adults with PWS can easily be taken advantage of by unscrupulous persons.

The right to contract

Adults with PWS typically lack the cognitive ability, judgment, and analytic skills necessary to understand contracts and therefore are not competent to enter into a legally binding agreement. Adults with PWS generally lack the ability to forecast potential options and weigh potential consequences or outcomes to actions. Adults with PWS can easily be taken advantage of or defrauded by unscrupulous persons.

The right to give or withhold medical consent

There are myriad serious and potentially life-threatening physiological symptoms caused by PWS. Adults with PWS generally lack adequate body awareness in order to assess the severity of an illness or injury. Most adults have a very high tolerance for pain; someone may walk for days on a broken leg or misidentify which leg is actually broken. The brain does not regulate the body's temperature properly; someone may be seriously, even life-threateningly ill but have no fever. At the same time, adults with PWS can feign illness or injury in order to receive sympathetic attention, or pursue hospitalization if they believe they can or they have a history of obtaining food. Adults with PWS lack the ability to weigh potential consequences or outcome to their actions or to forecast healthy potential options; disappointed or upset adults with PWS have abruptly terminated appropriate medical treatment and discharged themselves against medical advice.

The right to control social and sexual contacts and relations

Adults with PWS generally crave social connection and acceptance, however most adults are naïve, gullible and susceptible to manipulation and/or being taken advantage of. Some adults with PWS have admitted they cannot say "no" or turn down another's sexual advances or requests for sex for fear of losing the friendship, while others have been known to prostitute themselves for food or trade sexual behaviors for food or other wanted items.

The right to make decisions regarding education

Some adults with PWS love educational settings and do very well academically. Others may love the *idea* of higher education or specialized training but may not be able to emotionally manage the stress of the demands. It is extremely rare for an adult with PWS to safely operate a motor vehicle, and taking public transportation including a driving service such as Uber or Lyft can be problematic unless there is supervision to restrict access to unauthorized food or stops along the route to obtain food. It is imperative that adults with PWS be supervised when in the community, including any educational and specialized training environment, so as to restrict access to unauthorized food. Even the brightest, most articulate adult with PWS cannot always control the PWS hyperphagia symptom and therefore is at high risk of food-related death.

Do adults with PWS need *all* of their rights conserved?

PWS is a spectrum disorder, so while almost all of its symptoms are experienced by everyone with the diagnosis, not all symptoms are experienced to the same degree. Consequently, not everyone with PWS will need the same degree of conservatorship support. The individual's family or care providers will be your best resource to help determine which rights are necessary to ensure the health and safety of the adult with PWS.

It is important to remember that some persons with PWS present extremely well and appear far more capable than they actually are, however PWS specialists remind us that judgment is always impaired. For example, one may be able to dress oneself but not be able to choose clothing appropriate for the weather; or an academically-achieving student may consistently fail to look both ways before crossing the street; or a verbally articulate adult may actually be repeating phrases or ideas heard elsewhere as opposed to having original or insightful ideas; or an individual may believe if they drink or eat something they will become sick or even die but consume the item anyway. The lack of judgment exhibited by even the brightest or most articulate persons with PWS can be deadly.

Most people with PWS exhibit "delusional grandiosity" wherein they describe their abilities as far greater than they are or describe their symptoms as far less debilitating than they are. For example, someone may describe themselves as being a skilled swimmer but actually lack the ability to even tread water. Most often individuals are not intentionally lying; this behavior is simply a symptom of the syndrome.

In a nationwide survey conducted in 2010 of parents and care providers who have legal conservatorship of an adult with Prader-Willi syndrome, conservatorship exists over:

1. The right to fix the residence…….ALL
2. The right to access confidential records and papers……..…….....ALL
3. The right to consent to marriage or a registered domestic partnership.….MOST
4. The right to contract ……ALL
5. The right to give or withhold medical consent.……ALL
6. The right to control social and sexual contacts and relations……MOST
7. The right to make decisions regarding education…….....ALL

Who can I contact if I need more information about how PWS may affect the adult I've been asked to represent?

For more information about PWS and how it may affect the individual and/or family you are working with, contact the Prader-Willi California Foundation (PWCF) or the national Prader-Willi Syndrome Association (USA).

PWCF | 310-372-5053 | 800-400-9994 in CA | info@pwcf.org | www.PWCF.org

PWSA (USA) | 800-926-4797 | info@pwsausa.org | www.pwsausa.org

This document was created by PWCF's Home and Community-Based Services Task Force. Contact the PWCF Office to reach a member of the Task Force or if you need more information or support.

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Prader-Willi Syndrome: A Synopsis

Prader-Willi syndrome (PWS) (pronounced PRAH-der-WILL-ee) is a genetic disorder first identified in 1956 by Swiss doctors Prader, Willi and Labhart. Although PWS is generally associated with an abnormality of chromosome 15, there is no known cause for the genetic defect that results in this lifelong, life-threatening condition.

PWS is estimated to occur randomly in 1 in 10,000 to 15,000 people. The two most common types of PWS are Deletion and Uniparental Disomy (UPD). In almost all cases, neither type is inherited however genetic testing is recommended if more children are desired.

PWS is the most common genetic cause of obesity, and one of the ten most common conditions seen in genetics clinics.

CLINICAL FEATURES

First Stage—Failure to Thrive

Low birth weight and subsequent failure to thrive, severe muscle weakness (hypotonia), and delayed developmental milestones characterize the initial stage. Because of the weak muscle tone, motor planning deficits and swallowing issues, infants are often unable to nurse or suck and require special feeding techniques. The milestones of lifting the head, sitting up, crawling, and walking tend to be delayed. On average, without growth hormone treatment, independent sitting is achieved at around 12-13 months, walking at 24-30 months, and tricycle riding at 4 years. Without oral motor/speech therapy, the first words typically appear at around 21 months, with sentences at around 3½ years.

Second Stage—Hyperphagia

The next stage usually begins in toddlerhood when a preoccupation with food and the hyperphagia drive to eat begin. Life becomes dominated by a voracious appetite and an unrelenting drive for food. People with PWS do not experience normal satiety and can eat a tremendous amount of food without feeling ill. In addition to the drive for food, metabolism is at almost half the normal rate. Thus, if the food environment is not controlled the individual with PWS will quickly become obese.

CHARACTERISTICS

Abnormal growth: There is a deficiency in the production or utilization of growth hormone. Unless treated with growth hormone medication, persons with PWS are typically short in stature, have small hands and feet, and other subtle dysmorphic facial features.

Anxiety: Almost all persons with PWS experience high levels of anxiety. It is vital to reduce the stressors in the environment to the extent possible to reduce behavioral problems.

Behavioral challenges: Biochemical abnormalities in the brain cause increased anxiety and difficulty managing feelings of frustration and anger resulting in disruptive behavioral symptoms including Autism symptoms and yelling, verbal and physical aggression, and property destruction.

Body temperature regulation: The body's ability to regulate internal body temperature is often impaired. Most persons with PWS won't have an elevated temperature despite illness.

Cognitive limitations: IQs range from 40 to 100. Despite IQ, most persons with PWS have impaired judgment. Many individuals have learning disabilities, including Nonverbal Learning Disorder.

Dental problems: PWS causes low saliva production which results in thick, sticky saliva, soft tooth enamel, cavities, and gum disease. Products designed to treat dry mouth can improve or eliminate these symptoms.

Gastric & Bowel Problems: Gastroparesis (slow emptying stomach) and a slow emptying bowel are common. Often there is an inability to vomit despite severe illness. Stomach perforation or stomach tissue necrosis can result if there is unrestricted access to food.

High pain threshold: Most persons with PWS have a high tolerance for pain and are often unaware of injury or infection.

Hyperphagia: Persons with PWS experience a life-threatening biochemical drive to eat that is not satiated despite the quantity of food eaten. No known medication reduces or eliminates the hyperphagia food drive. Treatment currently consists of restricted access to food and continuous supervision throughout the lifetime.

Hypotonia: Infants generally exhibit severe muscle weakness. Muscle tone improves as the child ages but individuals never develop normal muscle strength and often fatigue easily.

Incomplete sexual development: Babies are typically born with small genitalia. Male babies are often born with undescended testes. Without sex hormone treatment, most adolescents do not produce sufficient sex hormones to progress through puberty.

Orthopedic Issues: Scoliosis, kyphosis, and other orthopedic abnormalities are common.

Psychiatric issues: There is a higher rate of psychiatric problems in persons with PWS than seen in the general population.

Metabolic issues: Metabolism is about half the normal rate therefore people with PWS will gain a considerable amount of weight on considerably fewer calories than the typical population. Fat tends to accumulate on the lower torso, buttocks, hips, thighs, and abdomen. Uncontrolled obesity can lead to high blood pressure, respiratory difficulties, heart disease, diabetes, and death. Caloric intake must be significantly reduced while preserving adequate nutrition. Consultation with a dietitian knowledgeable about PWS is recommended.

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Respiratory issues: Respiratory problems, including obstructive and central sleep apnea, can increase excessive daytime sleepiness and exacerbate behavior problems.

Skin picking: Persons with PWS often pick at their skin which, if not controlled, may result in infection.

Social isolation: Difficulties with social skills may lead to social isolation. Early Intervention programs should include social skills therapy. Individuals with higher levels of insight may be more aware of their differences or limitations which can lead to depression. Parents and care providers often experience social isolation.

Speech and language problems: Hypotonia and intellectual ability affect speech and language. Apraxia of Speech, also known as dyspraxia, is common. Oral-motor therapy in infancy and therapies targeted to treat dyspraxia are recommended.

Strength, balance and coordination: Along with the physical challenges caused by weak muscle tone, underdeveloped vestibular and proprioceptive systems often result in poor balance and coordination. Occupational and physical therapy are recommended to improve balance, coordination, and strength.

Swallowing abnormalities: Newer research reveals a high likelihood of swallowing problems that increase risk for choking and aspiration (fluid in the lungs).

Other characteristics: Sensitivity to medications, especially anesthesia; disordered sleep; eye abnormalities including strabismus (crossed eyes), myopia (nearsightedness), or amblyopia (lazy eye); lying, confabulating (making up) stories especially for food or attention; stealing food or money or impulsively stealing desired items; elopement.

TREATMENT

There is no cure for PWS—yet. There are, however, various therapies and treatment strategies that can help manage or reduce some of the symptoms… And there is hope.

Early Diagnosis and Therapies

Genetic testing is now widely used to diagnose PWS. Early diagnosis gives parents the opportunity to begin critical therapies and interventions as early as possible including proper nutritional intake, growth and sex hormone treatment, occupational and physical therapy, oral motor and speech and language therapy, sensory integration therapy, social skills therapy, and behavior therapy.

Growth Hormone Therapy

Growth hormone treatment in children with PWS is now considered the standard of care, and most PWS experts will prescribe growth hormone therapy for infants as well as adults. In addition to improved linear growth in children, the benefits of growth hormone therapy include improved muscle tone, improved respiratory function, improved cognitive function, decreased body fat, improved body composition, increased bone mineral density, improved physical performance, and, if administered prior to age 1, more normalized facial features.

Weight Control

While a hallmark symptom of PWS is the insatiable, unrelenting drive to obtain food, *no one with PWS is destined to become obese.* With proper nutrition, physical activity, and supervision people with PWS can maintain a healthy weight. Unfortunately, no medication currently exists to eliminate or even reduce hyperphagia, so access to food *must* be strictly controlled every day, every moment of the day. Providing necessary nutrients while restricting calories is essential. Exercise must begin early and be frequent and regular. Weight management should be individualized and include weekly weigh-ins and environmental barriers to food such as locks on the refrigerator and food pantries, no access to money that could purchase food, and continuous supervision to ensure absolute control of food intake.

Behavior Management

Implementing PWS-specific behavior management strategies and interventions as early as possible is crucial. These include but are not limited to establishing structure and daily routines, anticipating and preparing for any changes in the schedule, implementing the Principles of Food Security, and utilizing collaborative problem solving.

Special Education Services

Federal law requires states to provide services to handicapped children, including children with PWS. Services may include special instruction, occupational therapy, physical therapy, speech therapy, social skills therapy, psychological testing and counseling.

Specialized Residential Services

Until such time as there is successful treatment of the PWS hyperphagia food drive, persons with PWS need to live and work in settings that are trained to manage the unique, complex physiological and emotional symptoms caused by PWS. No one known to be diagnosed with PWS has successfully lived independently. Those who have tried have become morbidly obese and had to return to supervised residential setting or have died.

The outlook for persons diagnosed with PWS is more hopeful today than ever before. With strict food control and continuous supervision, the person with PWS may have a normal life expectancy and can accomplish many of the things their "typical" peers do — attend school, enjoy community activities, even move away from the family home to a supervised, structured residential setting. Research offers hope for new medications and treatments to better manage the multitude of symptoms caused by this complex disorder. To learn more about PWS contact the Prader-Willi California Foundation or the national Prader-Willi Syndrome Association (USA).

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