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 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 engaging in 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.

The outlook for persons diagnosed with PWS is more hopeful today than ever before. With strict food control, 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, work, even move away from home to an appropriate, supervised and 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 and how you can help, call us or visit our website.

Prader-Willi California Foundation is a non-profit 501(c)(3) corporation established in 1979. An affiliate of the Prader-Willi Syndrome Association | USA, PWCF is dedicated to supporting individuals with Prader-Willi syndrome, their families, and the professionals who serve them.

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A Synopsis

Prader-Willi Syndrome

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

Prader-Willi Syndrome

HISTORY

Prader-Willi syndrome (PWS) 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

Many of the symptoms of PWS can be reduced, managed or even eliminated. Contact PWCF or the PWSA (USA) for treatment and management guidance.

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 changes in the brain often cause increased anxiety and difficulty managing feelings of frustration and anger, often resulting in disruptive behavioral symptoms similar to Autism.

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, such as Biotene toothpaste, 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.

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.

Temperament and behavior issues: Young children with PWS tend to be happy and compliant. Over time due to biochemical changes in the brain, subtle changes may occur that cause the child to become more easily frustrated and prone to temper outbursts. There is a higher rate of psychiatric problems 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.

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.