

Fact Sheet: Prader-Willi Syndrome (PWS)

What is Prader-Willi Syndrome?

- A disorder of chromosome 15
- Prevalence: 1:12,000- 15,000 (both sexes, all races)
- Major characteristics: hypotonia, hypogonadism, hyperphagia, cognitive impairment, difficult behaviors
- Major medical concern: morbid obesity

Cause and Diagnosis of PWS

The genetic cause is loss of yet unidentified genes normally contributed by the father. Occurs from three main genetic errors: Approximately 70% of cases have a non-inherited deletion in the paternally contributed chromosome 15; approximately 25% have maternal uniparental disomy (UPD)—two maternal 15s and no paternal chromosome 15; and 2–5 % have an error in the "imprinting" process that renders the paternal contribution nonfunctional.

Diagnostic testing: Individuals who have a number of the clinical findings should be referred for genetic testing.

Major Clinical Findings

The following common characteristics of individuals with PWS raise suspicion of the diagnosis. Published diagnostic criteria include supportive findings and a scoring system ([Holm et al, *Pediatrics* 91, 2, 1993](#)).

- Decreased fetal movement, infantile lethargy, weak cry
- Characteristic behavior problems—temper tantrums, violent outbursts, obsessive/compulsive behavior; tendency to be argumentative, oppositional, rigid, manipulative, possessive, and stubborn; perseverating, stealing, lying
- Sleep disturbance or sleep apnea
- Short stature for genetic background by age 15
- Hypopigmentation—fair skin and hair compared with family
- Small hands and/or feet for height age
- Narrow hands with straight ulnar border
- Eye abnormalities (esotropia, myopia)
- Thick, viscous saliva with crusting at corners of the mouth
- Speech articulation defects
- Skin picking

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Weight and Behavior

Appetite Disorder

Compulsive eating and obsession with food usually begin before age 6. The urge to eat is physiological and overwhelming; it is difficult to control and requires constant vigilance.

Weight Management Challenge

Compounding the pressure of excessive appetite is a decreased calorie utilization in those with PWS (typically 1,000-1,200 kcal per day for adults), due to low muscle mass and inactivity. A balanced, low-calorie diet with vitamin and calcium supplementation is recommended. Regular weigh-ins and periodic diet review are needed. The best meal and snack plan is one the family or caregiver is able to apply routinely and consistently. Weight control depends on external food restriction and may require locking the kitchen and food storage areas. Daily exercise (at least 30 minutes) also is essential for weight control and health.

To date, no medication or surgical intervention has been found that would eliminate the need for strict dieting and supervision around food. GH treatment, because it increases muscle mass and function, may allow a higher daily calorie level.

Behavior Issues

Infants and young children with PWS are typically happy and loving, and exhibit few behavior problems. Most older children and adults with PWS, however, do have difficulties with behavior regulation, manifested as difficulties with transitions and unanticipated changes. Onset of behavioral symptoms usually coincides with onset of hyperphagia (although not all problem behaviors are food-related), and difficulties peak in adolescence or early adulthood. Daily routines and structure, firm rules and limits, "time out," and positive rewards work best for behavior management. Psychotropic medications—particularly serotonin reuptake inhibitors, such as fluoxetine and sertraline—are beneficial in treating obsessive-compulsive (OCD) symptoms, perseveration, and mood swings. Depression in adults is not uncommon. Psychotic episodes occur rarely.

Developmental Concerns

Motor Skills

Motor milestones are typically delayed one to two years. Proficiency with jigsaw puzzles is frequently reported, reflecting strong visual-perceptual skills.

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Oral Motor and Speech

Hypotonia may create feeding problems, poor oral-motor skills, and delayed speech. The need for speech therapy should be assessed in infancy. Sign language and picture communication boards can be used to reduce frustration and aid communication. Products to increase saliva may help articulation problems. Social skills training can improve pragmatic language use. Even with delays, verbal ability often becomes an area of strength for children with PWS. In rare cases, speech is severely affected.

Cognition

IQs range from 40 to 105, with an average of 70. Those with normal IQs typically have learning disabilities. Problem areas may include attention, short-term auditory memory, and abstract thinking. Common strengths include long-term memory, reading ability, and receptive language. Early infant stimulation should be encouraged and the need for special education services and supports assessed in preschool and beyond.

Growth

Failure to thrive in infancy may necessitate tube feeding. Infants should be closely monitored for adequate calorie intake and appropriate weight gain. Growth hormone is typically deficient, causing short stature, lack of pubertal growth spurt, and a high body fat ratio, even in those with normal weight. The need for GH therapy should be assessed in both children and adults.

Sexual Development

Sex hormone levels (testosterone and estrogen) are typically low. Cryptorchidism in male infants may require surgery. Both sexes have good response to treatment for hormone deficiencies, although side effects have been reported. Early pubic hair is common, but puberty is usually late in onset and incomplete. Fertility has not been documented in either sex.

Quality of Life Issues

General health is usually good in individuals with PWS. If weight is controlled, life expectancy may be normal, and the individual's health and functioning can be maximized.

The constant need for food restriction and behavior management may be stressful for family members. PWSA (USA) can provide information and support. Family counseling may also be needed.

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Adolescents and adults with PWS can function well in group and supported living programs, if the necessary diet control and structured environment are provided. Employment in sheltered workshops and other highly structured and supervised settings is successful for many. Residential and vocational providers must be fully informed regarding management of PWS.

Information from: Basic Facts About PWS: (amended from Prader-Willi Syndrome Association USA fact sheet) Diagnosis and Reference Guide for Physicians and Other Health Professionals