

Prader-Willi Syndrome

MEDICAL ALERT:

**A Diagnosis and Reference Guide for
Physicians and Other Health
Professionals**

Prader-Willi Syndrome

Basic Facts

5. DIAGNOSIS of infants with PWS is difficult. Newborns are hypotonic, lethargic, exhibit genital hypoplasia, and often require gavage or other special feeding techniques. Prenatally, fetal movement is decreased. Diagnosis in adults is more certain if classic characteristics are present: short stature, small hands and feet, CNS dysfunction, underdeveloped sexual characteristics, poor large muscle strength. Diagnostic criteria have been established (Holm, et. al: *Pediatrics* 01:398-402, 1993)

1. PRADER-WILLI SYNDROME (PWS) is an uncommon non-inherited birth defect, lifelong and life-threatening, affecting all races and both sexes. Prevalence is estimated to be 1:12,000 to 15,000.

6. MOTOR DEVELOPMENT is delayed typically on to two years, as are most milestones. For example, walking usually occurs around age two. Poor gross motor performance and balance improve slowly and lag behind normal individuals of the same age.

2. CHARACTERISTICS include hypotonia, insatiable appetite, obesity if food intake is uncontrolled, hypogonadism and incomplete sexual development, developmental delays, variable degrees of mental retardation or functional retardation, short stature (adult), small hands and feet, mild dysmorphism, and behavior problems which can be severe.

7. SPEECH AND LANGUAGE PROBLEMS are common. Cause is unclear, possibly poor muscle tone affecting vocal muscles and/or decreased saliva production. Speech therapy is recommended to relieve frustration associated with non-communication. Though language development is delayed, verbal ability is often a strength. Articulation may remain poor.

3. CAUSE is unclear. Approximately 70% have an interstitial deletion or other abnormal finding on chromosome 15, using high resolution (prometaphase) analysis. The remainder have two maternal chromosome 15s and no paternal 15 (maternal uniparental disomy).

8. AVERAGE I.Q. is around 70, with a range from 40 to 105. Typically, functioning is below I.Q. level. Abstract thinking and concepts are weaknesses. Even with a lower I.Q., cleverness in food seeking can exist.

4. RISK of recurrence in a family is low, estimated to be less than 1%.

9. BEHAVIOR PROBLEMS, ranging from stubbornness to violent temper tantrums and increasing with age, usually begin to appear during the preschool years when most are pleasant and cooperative. Intervention therapy can be used to modify severity (e.g., fluoxetine serotonin uptake inhibitors have been particularly beneficial in some cases). True depression and psychotic episodes are reported.

<p>10. COMPULSIVE EATING AND OBSESSION WITH FOOD begin usually between ages 2 - 4, though they may start later. Some learn to eat at fixed times and may refuse certain foods, but the insatiable drive for food persists. Sneaking or stealing extra food is common. In most situations, all sources of food must be kept under lock and key. Avoidance of temptation is helpful.</p>	<p>15. CONDITIONS ASSOCIATED WITH PWS include strabismus, myopia, scoliosis (may occur unusually early; often not recognized due to obesity), and diabetes (Type II, probably secondary to the obesity, responds well to weight loss and diet).</p>
<p>11. OBESITY occurs in 95% if there are no environmental controls. An increasing number are being diagnosed before onset of obesity. Whether obese at diagnosis or not, more and more have their weight managed within acceptable levels. Calorie utilization is decreased and diets should offer fewer calories than usual (often 1,000-1,200 per day). Exercise is critical to weight control.</p>	<p>16. MEDICATIONS, e.g., anorexic agents or psychoactive drugs, in most cases have not been found generally useful to control appetite. Drug treatment is essential at times but is not recommended for continuing usage. Consult the national PWSA office for updates. For some medications, increased sensitivity to normal doses is found.</p>
<p>12. SEXUAL DEVELOPMENT is deficient due to hypogonadotropic hypogonadism. Males have hypoplastic scrotum and cryptorchidism; females have hypoplastic labia minora and clitoris. Puberty may be early or late in onset and is usually incomplete. Females generally have oligomenorrhea or amenorrhea. Fertility has not been documented in either sex.</p>	<p>17. ADOLESCENTS AND ADULTS can function well in group living programs if they have adequate calorie control and structured living. Their rigid personalities and explosive tempers make supervision and programming difficult. Sheltered workshops generally provide the best employment possibilities.</p>
<p>13. SPORTS ACTIVITIES are limited. Running and jumping can cause joint injuries due to poor muscle strength and poor coordination. Incidence of bone fractures is increased, probably related to osteoporosis and decreased muscle bulk. Adaptive physical exercise is necessary in school. Walking, swimming, and stationary exercise equipment are recommended.</p>	<p>18. LIFE EXPECTANCY may be normal if weight is controlled. General health is usually excellent, although premature aging is reported. Obesity and its complications are the usual causes of morbidity and mortality.</p>
<p>14. DENTAL PROBLEMS are common and may include soft tooth enamel, thick sticky saliva, poor oral hygiene, sometimes teeth grinding, and infrequently rumination. Orthodontia must take into consideration delayed bone growth and abnormal puberty.</p>	<p>19. STRESS ON FAMILIES is often extreme. The constant pressure of food control and behavior management affects all family members, e.g., higher divorce rates and siblings with emotional problems. The child with PWS can become a tyrant who rules the family and, with age, becomes more difficult to handle. Awareness of these stresses and dealing with them early on is essential to the family structure.</p>

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CHARACTERISTICS OF THE SYNDROME THAT COULD CREATE COMPLICATIONS DURING ACUTE OR ROUTINE MEDICAL CARE, OR HOSPITALIZATION ARE LISTED BELOW:

1.) GENERALIZED RESPIRATORY DISTRESS or SLEEP APNEA

Due to massive obesity and hypoventilation syndrome. Excessive sleepiness is common. Generally occurs in the more obese, but also in normal weight persons with PWS. Weight loss can alleviate respiratory distress and may improve sleep disturbance.

2.) ADVERSE REACTION TO MANY DRUGS

Especially medication for weight reduction or behavior control. Prolonged sedation results from usual doses of IM drugs.

3.) HIGH PAIN THRESHOLD AND A DYSFUNCTIONING THERMOSTAT

Potential factors in risk of undetected infections. Many will not complain of pain until infection is severe. Idiopathic hyper- or hypothermia have been reported.

4.) LACK OF VOMITING

In about 2/3. Some will ingest almost anything (e.g., garbage). Emetics may be ineffective, and repeated doses may cause more problems.

5.) SCRATCHING and SKIN PICKING

Common, with scars and sores in various stages of healing. Sores often kept irritated for months by picking and may become infected.

6.) HYPOTONIA

Central, with poor suck and lethargy. Lasts for first several months of life. This leads to feeding problems, easy fatigue, failure to thrive and sometimes in difficulty clearing secretions.

7.) HYPOGONADISM

Underdeveloped genitals usually present throughout life. Cryptorchidism and inguinal

hernias are frequent in males. Inadequate secondary sex characteristics in both sexes have good response to hormone treatments, although side effects have been reported. Growth hormone has improved short stature due to lack of pubertal growth spurt and probable partial growth hormone deficiency and has increased lean body mass and resulted in decreased weight.


8.) HYPERPHAGIA

Insatiable appetite plus decreased resting metabolic rate lead to life-threatening weight gain, which can be very rapid and occur even on a low calorie diet. A typical diet still may contain too many calories. A nutritionally sound, restricted calorie diet is essential from early childhood.

Even normal weight persons with PWS cannot be trusted around food. All become masters at sneaking food around age 4. This skill increases with age. If weight is normal, it is because the caretaker, not the person with PWS, has achieved control via such steps as locks in the kitchen, constant vigilance, and adequate exercise/activity. It is very hard to have food visible or be around others who are eating all they want. To date, no medication or surgical intervention has been found helpful in most cases for long-term weight control.

9.) BEHAVIOR and EMOTIONAL PROBLEMS

Due to the CNS dysfunction, complicated by multiple factors, most children and adults with PWS have difficulty with stubbornness, temper tantrums, and depression, all of which increase with age. When persons with PWS are distraught, "talking over" the problem generally makes them more upset. Firm limits, "time out", and positive rewards work best for behavior management. Medications may help short term. Serotonin uptake inhibitors help in some cases in usual doses or less.



The Prader-Willi Syndrome Association (USA) was formed in 1975 in order to provide a vehicle for communication for parents, professionals and other interested citizens. It is an organization dedicated to the sharing of experiences in how to cope with the syndrome. Chapters of PWSA(USA) are located in most states and are available for support, education and advocacy.

An annual national conference, a bi-monthly newsletter entitled *The Gathered View*, which incorporates contributions from members as well as professional advice and a wealth of other written materials and publications on Prader-Willi Syndrome, are all part of PWSA's effort to carry out its mission.

Supported solely by membership fees and donations, Prader-Willi Syndrome Association (USA) has made a difference in the lives of many affected by this unique syndrome. YOU, TOO, CAN HELP. Donations and membership applications may be sent to the address below.

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