

PRADER-WILLI SYNDROME (PWS)

(pronounced "Praw-der" - "Wil-lee")

Prader-Willi Syndrome (PWS) is a rare genetic disorder that occurs in approximately 1 out of every 12,000 - 15,000 births. It is the most common genetic cause of life-threatening obesity, affecting appetite, growth, metabolism, cognitive functioning and behavior. PWS is a lifelong condition in which there is no known cure... YET!



TWO STAGES OF SYMPTOMS:

1) INFANCY STAGE

- **Severe Muscle Weakness** (arms and legs are "floppy"). Muscle tone is affected throughout entire body (mouth, throat, lungs, heart, etc.). Strength and tone generally improve with age & help of growth hormone treatment.
- **Weak Cry & Poor Suckle** Babies sleep a lot, do not cry very much, if at all, and often need to be woken up to eat. They typically are unable to feed from a bottle and often need a feeding tube to eat.
- **Sleep Apnea** Some babies require oxygen because their brains don't tell them to breathe regularly.
- **"Failure to Thrive"** Babies may not gain weight or grow normally; may occur if feeding problems aren't monitored closely.
- **Delayed Motor/Physical Milestones** Skills like rolling over, sitting up, crawling, standing, walking, speech – these are eventually achieved, but later in infant/child's life with intervention of physical, occupational & speech therapies.



2) HUNGER STAGE

- **Uncontrollable Appetite** (typically begins between the ages of 1 - 6 years old). People with PWS are not able to control their food intake and will overeat if not closely monitored. Part of their brain (hypothalamus) does not tell them when they are "full" after eating, so they *always* feel hungry. Some people with PWS will do anything to find food to eat. For this reason, refrigerators and food cabinets may need to be locked in their homes.
- **Slow Metabolic Rate** – runs approximately 60% of normal. People with PWS do not burn calories as easily as others. This makes it much harder for them to lose weight.
- **Low Energy Level** It's difficult to exercise or play sports as they will tire easily.

PWSA (USA)

8588 Potter Park Dr., Ste. 500

Sarasota, FL 34238

(800) 926-4797 ▲ www.pwsausa.org

Insert your local/state chapter info

Prader-Willi Syndrome of...

[www.\(yourwebsite\).org](http://www.(yourwebsite).org)

Call (---) 555-0000 or

Email: pws--@--.com

Other Symptoms/Characteristics:

- Small Hands & Feet
- Short Stature
- Down-turned Mouth
- Delayed Speech
- Behavior Challenges
- Scoliosis
- High Pain Threshold
- Temperature Dysregulation
- Skin Picking
- Bruise Easily
- Breathing Problems
- Lack of Vomiting
- Dental Problems
- Strabismus
- Hypopigmentation



PRADER-WILLI SYNDROME ASSOCIATION
— Still hungry for a cure. —

Questions & Answers on Prader-Willi Syndrome (PWS)

Q: WHAT IS PWS?

A: PWS is a complex genetic disorder that typically causes low muscle tone, short stature, incomplete sexual development, cognitive disabilities, problem behaviors, and a chronic feeling of hunger that can lead to excessive eating and life-threatening obesity.

Q: IS PWS INHERITED?

A: Most cases of PWS are attributed to a spontaneous genetic error that occurs at or near the time of conception for unknown reasons. In a very small percentage of cases (2% or less), a genetic mutation that does not affect the parent is passed on to the child, and in these families more than one child may be affected. A PWS-like disorder can also be acquired after birth if the hypothalamus portion of the brain is damaged through injury or surgery.

Q: HOW COMMON IS PWS?

A: It is estimated that one in 12,000 to 15,000 people has PWS. Although considered a "rare" disorder, PWS is one of the most common conditions seen in genetics clinics and is the most common genetic cause of obesity that has been identified. PWS is found in people of both sexes and all races.

Q: HOW IS PWS DIAGNOSED?

A: Suspicion of the diagnosis is first assessed clinically, then confirmed by specialized genetic testing on a blood sample. Formal diagnostic criteria for the clinical recognition of PWS have been published (Holm et al., 1993), as have laboratory testing guidelines for PWS (ASHG, 1996).

Q: WHAT IS KNOWN ABOUT THE GENETIC ABNORMALITY?

A: Basically, the occurrence of PWS is due to lack of several genes on one of an individual's two chromosome 15s - the one normally contributed by the father. In the majority of cases, there is a *deletion* - the critical genes are somehow lost from

the chromosome (deletion). In most of the remaining cases, the entire chromosome from the father is missing and there are instead two chromosome 15s from the mother (uniparental disomy). The critical paternal genes lacking in people with PWS have a role in the regulation of appetite. This is an area of active research in a number of laboratories around the world, since understanding this defect may be very helpful not only to those with PWS but to understanding obesity in otherwise normal people.

Q: WHAT CAUSES THE APPETITE AND OBESITY PROBLEMS IN PWS?

A: People with PWS have a flaw in the hypothalamus part of their brain, which normally registers feelings of hunger and satiety. While the problem is not yet fully understood, it is apparent that people with this flaw never feel full; they have a continuous urge to eat that they cannot learn to control. To compound this problem, people with PWS need less food than their peers without the syndrome because their bodies have less muscle and tend to burn fewer calories.

Q: DOES THE OVEREATING ASSOCIATED WITH PWS BEGIN AT BIRTH?

A: No. In fact, newborns with PWS often cannot get enough nourishment because low muscle tone impairs their sucking ability. Many require special feeding techniques or tube feeding for several months after birth until muscle control improves. Sometime in the following years, usually before school age, children with PWS develop an intense interest in food and can quickly gain weight if calories are not restricted.

Q: DO DIET MEDICATIONS WORK FOR THE APPETITE CHALLENGES IN PWS?

A: Unfortunately, appetite suppressants haven't worked consistently for people with PWS. Instead, an extremely low-calorie diet is required, as well as an environment designed to limit access to food. For example, many families have to lock their kitchen cabinets and refrigerator.

As adults, some individuals find controlling their weight is more easily managed in a PWS group home setting, as food access can be monitored without interfering with those who don't require such restriction.

Q: WHAT KINDS OF BEHAVIORAL ISSUES DO PEOPLE WITH PWS HAVE?

A: In addition to their involuntary focus on food, people with PWS tend to have obsessive/compulsive behaviors that are not related to food. Some are repetitive thoughts and verbalizations, collecting and hoarding of possessions, picking at skin irritations, and a strong need for routine and predictability. Frustration or changes in plans can easily set off a loss of emotional control in someone with PWS, ranging from tears and temper tantrums to physical aggression. While psychotropic medications can help some individuals, the essential strategies for minimizing difficult behaviors in PWS are careful structuring of the person's environment and consistent use of positive behavior management and supports.

Q: DOES EARLY DIAGNOSIS HELP?

A: While there is no medical prevention or cure, early diagnosis of PWS gives parents time to learn about and prepare for the challenges that lie ahead. Establishing a family routine that will support their child's diet and behavioral needs from the start is vital to their success. Knowing the cause of their child's developmental delays can facilitate a family's access to important early intervention services and may help program staff identify areas of specific need or risk. Additionally, a diagnosis of PWS opens the doors to a network of information and support from professionals and other families who are dealing with the syndrome.

Info provided by...

PRADER-WILLI SYNDROME ASSOCIATION
—Still hungry for a cure.—