

# Prader-Willi Syndrome

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# What is Prader-Willi Syndrome?

- A disorder of Chromosome 15
- Occurs in 1 out of every 12,000-15,000 births (both genders, all races)
- Major characteristics: hypotonia, cognitive impairment, difficult behaviors
- Major medical concern: morbid obesity

Prader-Willi syndrome (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. (Prader-Willi Syndrome Association (USA) website)

# Is PWS inherited?

- Most cases of PWS are attributed to a spontaneous genetic error that occurs at or near the time of conception. The reason for this error is unknown. In 2 percent or less of the cases a genetic mutation that does not affect the parent is passed on, in these families more than one of the children may be affected. A PWS-like disorder can also be acquired after birth if there is damage caused to the hypothalamus portion of the brain due to injury or surgery.

# Symptoms of PWS

- Floppy newborn infant (hypotonic)
- Small for gestational age
- Undescended testicles for males
- Delayed motor development
- Slow mental development
- Very small hands and feet in comparison to body
- Rapid weight gain
- Food cravings
- Almond-shaped eyes
- Narrow bifrontal skull
- Morbid obesity
- Skeletal (limb) abnormalities

# How is PWS diagnosed?

- Doctors use a checklist of characteristics in the clinic setting if they are suspicious that a child has PWS. After they have confirmed their suspicions they send a blood sample in for specialized genetic testing for a formal diagnosis.

# Does an early diagnosis help?

- There is no cure for PWS, but early diagnosis can help a family to adapt healthy living habits. It can also help them to learn about and prepare for the challenges that are ahead.

# Does overeating begin at birth?

- No, most infants with PWS have low muscle tone and cannot get enough nourishment because it limits their sucking ability. Many require special feeding techniques for several months after birth.
- In the years to follow, usually before school age, the children develop an increased interest in food and can quickly gain weight.

# What causes the appetite and obesity problems in PWS?

- People with PWS have a flaw in the hypothalamus. (The hypothalamus usually tells the body whether it is hungry.) They never get the sense of feeling full and have a continuous urge to eat that they cannot learn to control.
- People with PWS need less food than their peers because they have less muscle and burn fewer calories.

# Are there any medications to help treat PWS?

- There is not an appetite suppressant that has worked consistently. Most require an extremely low-calorie diet all their lives and have an environment that gives them very limited access to food.
- Research is ongoing!

# Do people with PWS have behavior problems?

- People with PWS tend to have obsessive/compulsive behaviors that aren't related to food. For example: picking at their skin, a strong need for routine, collecting possessions, repetitive thoughts
- Some psychotropic medications can be used, but the best strategies for these behaviors is to carefully structure the person's environment.

# What does the future look like for people with PWS?

- People with PWS can expect to do many of the things that their “peers” do—complete school, become employed, move away from home.
- They do need support from their families and friends to achieve these goals and avoid serious health consequences.

# For more information...

- Contact the Prader-Willi Syndrome Association (USA) at 1-800-926-4797 or on the web at [www.pwsausa.org](http://www.pwsausa.org)
  - The PWSA (USA) offers a toll-free helpline, bimonthly member newsletter, a variety of additional publications, and annual family conference and scientific meeting, and chapters throughout the USA to provide family support and advocacy

# Resources

- Dictionary of Developmental Disabilities, Terminology, Second Edition, [www.brookespublishing.com/cgi-bin/dictionary.pl](http://www.brookespublishing.com/cgi-bin/dictionary.pl)
- Prader-Willi facts from The Arc, [www.thearc.com](http://www.thearc.com)
- Medline Plus Medical Encyclopedia, [www.nlm.nih.gov/medlineplus/ency/article/001605.htm](http://www.nlm.nih.gov/medlineplus/ency/article/001605.htm)
- Prader-Willi Alliance, [www.prader-willi.org](http://www.prader-willi.org)