



Rebecca

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, Pediatrics 91, 398,1993) and further refined (Gunay-Aygun M, Schwartz S, Heeger S, O’Riordan MA, Cassidy SB. The changing purpose of Prader-Willi syndrome clinical diagnostic criteria and proposed revised criteria. Pediatrics, 108(5):E92, 2001). The diagnostic criteria and also laboratory testing guidelines for PWS can be found on the PWSA (USA) website www.pwsausa.org, as well as in the 3rd Edition of our “Management of Prader-Willi Syndrome” textbook. Genetic testing is now very accurate for the diagnosis.

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.



Cameron

Q: What is Prader-Willi syndrome (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, IF FOOD ACCESS IS NOT RIGOROUSLY CONTROLLED, 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 and therefore is not inherited. In a very small percentage of cases (2 percent or less), a genetic defect that does not affect the parent can be 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 in 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 newborns has PWS. Although considered a “rare” disorder, Prader-Willi syndrome is one of the most common conditions seen in genetics clinics and is the most common genetic cause of life-threatening obesity that has been identified. PWS is found in people of both sexes and all races.

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

A national conference, a bimonthly 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 the PWSA (USA)’s effort to carry out its mission.

PWSA (USA) is supported solely by membership fees and donations. Our agency has made a difference in the lives of many families and individuals affected by this unique syndrome. **YOU TOO CAN HELP.** Donations and membership applications may be sent to the address below.

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We hope you find these materials helpful and that you consider a donation to PWSA (USA) to assist in developing more good work(s) like this. Please see our website, www.pwsausa.org.

Questions and Answers



Reagan

On Prader-Willi Syndrome

USA
PRADER-WILLI SYNDROME ASSOCIATION *Still hungry for a cure.*

Our Mission

Prader-Willi Syndrome Association (USA) is an organization of families and professionals working together to raise awareness, offer support, provide education and advocacy, and promote and fund research to enhance the quality of life of those affected by Prader-Willi syndrome.



Elena



Graisen



Addy

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 excess weight if calories are not restricted.

Q: Do diet medications work for the appetite problems in PWS?

A: Unfortunately, no appetite suppressant to date has worked consistently for people with PWS, but there are promising drugs in clinical trials. Most require an extremely low-calorie diet all their lives and must have their environments designed

so that they have very limited access to food. For example, many families have to lock the kitchen or the cabinets and refrigerator. As adults, most affected individuals can control their weight best in a supportive living home designed specifically for people with PWS, where food access can be restricted without interfering with the rights of those who don't need such restriction.

Q: What kinds of behavior problems 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, such as 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. 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 Prader-Willi syndrome gives parents time to learn about and prepare for the challenges that lie ahead and to establish family routines that will support their child's diet and behavior needs from the start. 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.

Q: How does growth hormone therapy help?

A: Use of growth hormone therapy is becoming standard care for those with PWS, when prescribed with appropriate precautions by an experienced endocrinologist. Growth hormone therapy can help with height, weight, body mass, strength, and agility, among other areas. It may also help with cognitive development. Reports have indicated dramatic increase in growth rate (especially in the first year of treatment) and a variety of other effects, including improved body composition (higher muscle mass, lower fat mass); improved weight management; increased energy and physical activity; improved strength, agility and endurance; and improved respiratory function.



Jacob

Q: What does the future hold for people with PWS?

A: With help, people with PWS can expect to accomplish many of the things their "normal" peers do - complete school, participate in their outside areas of interest, be a productive worker under the right conditions, even move away from their family home. They do, however, need a significant amount of support from their families and from school, work and residential service providers, both to achieve these goals and to avoid obesity and the serious health consequences that accompany it. Even those with IQs in the normal range need lifelong diet supervision and protection from food availability. Although in the past many people with PWS died in adolescence or young adulthood, prevention of obesity can offer those with the syndrome the probability of living a normal life span.

New medications, including psychotropic drugs and synthetic growth hormone, are already improving the quality of life for some people with PWS. Ongoing research offers the hope of new discoveries that will enable people affected by this unusual condition to live more independent lives.