

Prader-Willi Syndrome

Description

Prader-Willi Syndrome (PWS) is the most commonly known genetic cause of life-threatening obesity in children. This disorder is a neurobehavioral disorder that is of genetic origin. It results from an abnormality on the 15th chromosome. Most people affected are diagnosed through genetic testing in early childhood. However, there are cases where a person can “acquire” PWS-like symptoms. This is very rare and attributed to an injury to the hypothalamic region of the brain. These individuals may show all signs and characteristics of PWS but lack the genetic or other clinical confirmation of the diagnosis. Regardless of the cause, the supports needed for the individual remain the same.

PWS is a result of damage to an area of the brain called the hypothalamus. The altered functions of the hypothalamus can lead to decreased growth hormones, altered reproductive hormones, and altered regulation of the autonomic nervous system. Autonomic nervous system abnormalities could result in problems with regulating body temperature, fluid balance, and sleep patterns.

Characteristics

There are unique characteristics of people with PWS. These characteristics are inclusive of physical, emotional, and behavioral factors.

- **Hyperphagia:** Individuals with PWS do not display this characteristic in the same way. Damage to the hypothalamus results in the ingestion of a greater than optimal quantity of food, and this has significant health risks. Unmanaged hyperphagia can lead to the ingestion of non-food items, and this situation has the potential to be life threatening.
- **Excessive food-seeking behaviors:** Individuals with this syndrome can develop complex skills related to obtaining food. This can include lying, stealing, pre-planning, and manipulation.
- **Obsessive-Compulsive Behaviors:** It is essential to remember that these characteristics are not necessarily maladaptive behaviors that require intervention. Rather these characteristics may be positive coping strategies for the individual as a way to deal with the syndrome.
- **Physical Appearance:** For men, average height is 5’0”. Women with PWS have an average height of 4’11”. Both men and women have small hands and feet.
- **Sexual Development:** Individuals with PWS usually do not complete puberty. This may compromise normal development of sexuality. Men do not develop facial hair and their voices do not deepen. Women often do not have a menstrual cycle and are unable to have children.
- **Impulsivity/Aggression:** There is usually a reason behind each impulsive or aggressive behavior. This could often be attributed to impulsive food-seeking behavior and/or the frustration related to obsessively seeking food.

- **Moderate to Mild Intellectual Disabilities:** This may be deceptive, giving the impression that the individuals with this syndrome have the ability to control their actions.

Incidence and Prevalence

PWS affects approximately 1 in 12,000-15,000 newborns in the US. It is not inherited, but a chromosomal abnormality that occurs by chance. There is research to suggest the Imprinting Mutation form of PWS could have a chance of inheritance, and families with individuals affected by this form of PWS may benefit from genetic counseling. However, there is no conclusive research to suggest any form of prevention and or known cure for PWS.

Other Potential Issues and Factors

Individuals with PWS function best in a highly structured environment where access to food is controlled and monitored very closely. Individuals with PWS have:

- Low metabolism and inactivity. Paired with the brain's failure to tell them they are full, it is imperative that individuals with PWS stay on a lower calorie diet that will not lead to other serious health issues.
- Hypotonia, or weak muscle tone, can lead to an increase in a sedentary lifestyle further exacerbating the potential for life-threatening obesity.
- Sleep disturbances include decreased breathing with lower blood-oxygen levels during sleep.
- Unusual reactions to standard dosages of medication.
- Limited or no ability to feel pain. The research literature has conflicting findings. However, it would be beneficial to teach a daily routine of checking extremities for injuries, as one would instruct an individual with diabetes.
- Due to the excessive eating and the possible eating of uncooked, spoiled, or otherwise unhealthy food items, lack of vomiting is of particular health and safety concern.
- Potential for skin infections or lesions due to excessive skin and nose picking that may need to be managed as a life long concern. This is the most common form of self-injurious behavior observed in individuals with PWS.

Possible Interventions and Supports

There is no prevention or cure for PWS. Some individuals with PWS can lead independent lives, but some individuals will need life-long supervision to ensure their health and safety. A highly structured environment with appropriate modifications for safety can assist individuals with PWS to develop to their fullest potential. Strategies such as using visual schedules, pre-teaching and rehearsal techniques, and self-monitoring can assist with developing independence.

Many individuals with PWS desire companionship, and it is important for individuals with PWS to develop a network of natural supports. This is a team comprised of community supports, the faith community, family, and friends. A strong network of natural supports is essential to help the individual create a sense of self-determination, independence, opportunity, and support. This “team” can also be a true deterrent to exploitation.

Effective and important practices that will help an individual with PWS to thrive include:

- Following a low-calorie diet monitored by nutritionist specializing in PWS.
- Weighing and measuring food and drink intake.
- Monitoring weight closely and addressing any dramatic changes immediately.
- Teaching the ability to self-monitor.
- Teaching strategies for planning and accepting change.
- Exercising daily.
- Modifying environment.
- Using assistive technology to develop independence.
- Establishing routine and structure.
- Arranging physical set-up of kitchen, food storage, bedrooms, medication, personal hygiene supplies, money, privacy, alarms, and outside trash areas.
- Understanding what is motivating to the individual.
- Planning and scheduling for social events or any community-based activities where food is available
- Implementing formal behavior management and positive behavior supports.
- Teaching methods to deal with the frustration of not being able to feel fulfilled in their desires to seek food.
- Understanding the underlying reasons for any aggressive behaviors and teach positive pro-social coping strategies.
- Assisting individuals to develop their own natural supports system.
- Teaching skills to delay gratification and improve impulse control.
- Educating individuals to deal with challenges associated with this diagnosis.

Cultural Considerations

PWS is seen in all races; equally in both males and females. Symptoms of PWS are present at birth or acquired later from a brain injury and remain life-long. Most societies are extremely focused on food. This situation can be very stressful and life threatening for people with PWS. Regardless of culture, ethnicity, or religion, the focus of food related to traditions, holidays, social events, and activities of daily life is significant, so on-going planning is imperative to ensure the health and safety of the individual from the critical hazards of obesity.

References and Resources

Print Resources

Goff, B. J. (2008). *Supporting adults with prader-willi syndrome in residential settings*. Sarasota, FL: Prader-Willi Syndrome Association (USA).

Medical Specialists in Prader-Willi Syndrome. (2009) *Prader-Willi Syndrome medical alerts booklet*. Sarasota, FL: Prader-Willi Syndrome Association [USA].

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Prader-Willi Association [USA] (<http://pwsusa.org>)

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