Published on *My46* (https://www.my46.org) 01/20/2022

My46 Trait Profile

Prader-Willi syndrome

Other Names: Prader-Labhart Willi syndrome

Prader-Willi syndrome is a genetic condition characterized by poor muscle tone and feeding difficulties in infancy followed by excessive eating in early childhood. It is caused by the lack of expression of critical genes in a particular region of the paternal copy of chromosome 15.

Characteristics of Prader-Willi syndrome

Prader-Willi syndrome is a genetic condition that is typically characterized by low muscle tone, which often leads to reduced muscle strength, and feeding difficulties in early infancy followed by excessive food intake in early childhood. This excessive eating can lead to extreme obesity unless it is regulated and controlled. An underactive thyroid gland (i.e., hypothyroidism) is a common feature seen in Prader-Willi syndrome. Developmental delays include speech, gross and fine motor skills. All individuals with Prader-Willi syndrome are affected with some degree of cognitive impairment. There are distinct behavioral characteristics including temper-tantrums, stubbornness, and obsessive-compulsive tendencies. Reduced functionality of the sex organs, also known as hypogonadism, is present in both males and females. This usually leads to incomplete pubertal development and the inability to have children. Short stature, scoliosis, and strabismus, also known as cross-eyed or lazy eye, are also common. Individuals with Prader-Willi syndrome who become obese are at a higher risk of developing type II diabetes and high blood pressure.

Diagnosis/Testing

The diagnosis of Prader-Willi syndrome can be based upon clinical findings alone. Genetic testing is valuable for confirming the diagnosis and for providing genetic counseling. There are three primary genetic causes for Prader-Willi syndrome. Approximately 70-75% of individuals with Prader-Willi syndrome have a missing piece (deletion) on chromosome 15 that came from the father. Approximately 25% of individuals have Prader-Willi syndrome as a result of maternal uniparental disomy (i.e., when both copies of chromosome 15 are inherited from the mother instead of one copy from each parent). The least common cause of Prader-Willi syndrome is due to abnormal imprinting, or expression of only the maternal genetic information within a critical region on chromosome 15.

Management/Surveillance

During infancy, special nipples or feeding tubes should be utilized to ensure that the proper amount of nutrients is being consumed. Physical therapy is recommended to improve muscle strength. Beginning in infancy, individuals with Prader-Willi syndrome should have regular eye exams and be monitored frequently for height, weight, and BMI to assure that proper weight precautions are being taken. Appropriate and regular exercise regimens should be implemented. In addition, annual testing for hypothyroidism should be conducted. During childhood when food intake begins to drastically increase, consumption should be under strict supervision based on height, weight, and BMI (body mass index) in order to prevent excessive weight gain. Growth hormone replacement therapy can be utilized to normalize height, increase muscle tone and decrease fat mass. Educational therapies, specifically for language development, should begin as early as possible. Behavioral therapies in combination with behavioral psychotropic medications may be helpful for teens and adults with Prader-Willi syndrome. Through the use of sex hormones at

puberty, adequate secondary sexual characteristics can be achieved.

Mode of inheritance

Prader-Willi syndrome occurs in all populations around the world and affects girls and boys equally. Most cases of Prader-Willi syndrome are sporadic, meaning that there are no other family members with Prader-Willi syndrome.

Risk to family members

The chance for parents of a child with Prader-Willi syndrome to have another affected child depends on the genetic cause. Most deletions, uniparental disomy, and imprinting center defects occur brand new (de novo) and are not inherited. Thus, the risk of other siblings being affected is low. However, some genetic causes can be inherited from the father. For example, when a father is found to have the same deletion in the imprinting center found in his child with Prader-Willi syndrome, the risk of having another child with Prader-Willi syndrome is as high as 50% with each pregnancy.

Special considerations

None

Resources

Genetics Home Reference: Prader-Willi syndrome
http://ghr.nlm.nih.gov/condition/prader-willi-syndrome
University of Michigan Health System: Prader-Willi Syndrome
http://www.med.umich.edu/yourchild/topics/praders.htm
Prader-Willi Syndrome Association
http://www.pwsausa.org/index.html
Medical Home Portal: Prader-Willi Syndrome
http://www.medicalhomeportal.org/diagnoses-and-conditions/prader-willi-syndrome/description

References

Cassidy, SB. et al. (2012)."Prader-Willi syndrome." Genetics in Medicine 14(1): 10-26.
Chen, C. et al. (2007)."Prader-Willi syndrome: An Update and Review for the Primary Pediatrician." Clinical Pediatrics 46(7): 580-591.
Driscoll DJ, Miller JL, Schwartz S, Cassidy SB. (Updated 11 October 2012). Prader-Willi Syndrome. In: GeneReviews at GeneTests Medical Genetics Information Resource (database online). Copyright, University of Washington, Seattle. 1997-2013. Available at http://www.ncbi.nlm.nih.gov/books/NBK1330/. Accessed [11/01/2013].
McCandless, SE. et al. (2011)."Clinical report – health supervision for children with Prader-Willi syndrome." Pediatrics 127(1): 195-204.

Created: 11/2013 **Created by:** Daniela Iacoboni, MS, CGC
Updated: mm/yyyy **Edited by:** Seema Jamal, MSc, LCGC