

Prader-Willi Syndrome

Prader-Willi Syndrome is a collection of features and problems caused by genetic changes on chromosome 15. Prader-Willi Syndrome was first named in 1956. In 1981, Dr. David Ledbetter and colleagues found that many people with Prader-Willi Syndrome had the same segment of genes missing from one of their chromosomes. The deletion on chromosome 15 accounts for more than half of the cases of PWS.

Testing for PWS is complicated and it usually takes more than one test to be certain of a diagnosis of PWS. This birth defect affects about 1 in 15,000. It affects both races and sexes equally. There is no cure, only treatment.

Characteristics include short stature, mental retardation or learning disabilities, incomplete sexual development, low muscle tone and a constant urge to eat. Because they do not metabolize calories normally, they tend to be obese. They exhibit poor gross motor skills, speech and language difficulties, mood swings, behavior problems, cognitive limitations, high pain threshold, skin picking and dental problems.

Resources

Information obtained from the following web sites. www.pwsausa.org This is the Prader-Willi Syndrome Association (USA) site. This site has much more on the genetics of PWS

www.midnet.sc.edu/prader-willi This is the Prader-Willi Syndrome Association of South Carolina web site. Submitted by Penny Adsit.

