

Students with Prader-Willi Syndrome – AN OVERVIEW

NOTE: The following are excerpts from the publication, *The Student with Prader-Willi Syndrome: Information for Educators*. This publication in its entirety is available through PWSA (USA).

Students with Prader-Willi syndrome (PWS) are students you never forget. No matter what their age, they will teach you many things. Those that are very gifted in perseverance and ingenuity ... teach you patience and problem solving. Those that require structure and consistency ... teach you planning and organization. Those that are outgoing and friendly ... teach you warmth and laughter. Students with PWS are as much individuals as any students. Some of the information provided will be very applicable to the student you are working with; some will not. Some strategies will work; others will not. With knowledge and understanding, students with PWS become productive members of their communities.

A Brief Overview of Prader-Willi Syndrome

PWS is a complex neurobehavioral genetic disorder. It affects approximately 1 in 12,000 – 15,000 newborns. It results from an abnormality on the 15th chromosome. It is seen in all races; equally in both males and females. Symptoms of PWS are present at birth and remain life long. Currently, there is no cure. Research is underway to broaden our understanding and treatment options.

Because of the advances in genetic testing, it is believed that most affected persons can be diagnosed with this disorder. There are cases where an individual can “acquire” PWS as a result of damage to the hypothalamus. This may be seen in a person who has a brain tumor or suffers injury to that area of the brain. There are also situations where people show all of the signs and characteristics of PWS but lack genetic or other clinical confirmation. No matter the cause, support needed for these individuals remains the same.

Characteristics and Stages

A syndrome is a group of signs and symptoms that collectively may indicate a particular disorder or condition. In most cases, not all persons affected by a disorder have all of the symptoms and the symptoms may differ in degree and severity.

The first stage “FAILURE TO THRIVE”	The second stage “THRIVING TOO WELL”
Birth to between 2 to 5 years.	Between 2-5 years throughout lifetime
<ul style="list-style-type: none"> • Low birth weight, decreased activity in mother’s womb; babies often arrive past their due date. • Weak muscle tone – “floppy baby.” • Feeding difficulties - poor sucking reflexes. Babies often require special feeding techniques. • Poor weight gain and slow physical growth. • Small hands and feet. • Excessive sleepiness. • Developmental delays. • Speech and language difficulties and delays. • Underdeveloped genitals – undescended testicles in boys. 	<ul style="list-style-type: none"> • Increased appetite and weight control problems. May begin as food preoccupation: advancing to food seeking and stealing in varying degrees and rates. Rapid weight gain if food and nutrition is not closely monitored. • Behavior problems. May see difficulty and inability to handle changes without becoming greatly upset. Temper tantrums and stubbornness can be severe. • Learning difficulties with approximately 60% having mental retardation/cognitive disability. • Speech and language difficulties. Varying expressive language problems. In many cases, receptive language is minimally affected. • Fine and gross motor delays. • Short stature if growth hormone therapy not initiated. • Social skills deficit.

Genetics and Diagnosis in PWS

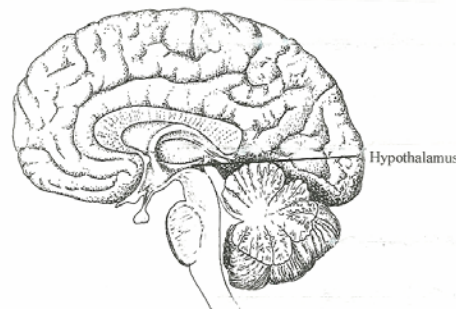
PWS results from an infant not receiving critical genes from a specific section of chromosome 15 that comes from the father. This happens for no reason. The risk of reoccurrence is low. There is nothing either parent did to cause it. Genetic counseling is advised. There are three different ways this can happen. The following descriptions are very brief.

1. **Paternal deletion** occurs in approximately 70% of cases. It is the most common form. Part of the chromosome inherited from the child's father – the part containing the PWS imprinted genes – is missing or broken.
2. **Maternal Uniparental Disomy (UPD)** occurs in approximately 25 % of cases. In this form of PWS, a child inherits both copies of chromosome 15 from the mother.
3. **Imprinting mutation** occurs in less than 5% of cases. In rare cases, the PWS genes on the father's chromosome are present but do not work because the process that normally switches them "on" does not work. If a person with PWS has an imprinting mutation, genetic counseling is strongly recommended. This type of mutation may be seen in siblings and other family members. Any child or adult that has a number of characteristics that were noted on the chart enclosed should be referred to a geneticist for genetic testing.

PWS and the Hypothalamus

PWS impacts the functioning of the hypothalamus. This area of the brain is responsible for many biological functions. It has many nerve connections with various parts of the body. It responds to changes in blood properties including pH, fluid balance and glucose levels. Its primary purpose is to maintain balance in our body. Many hormones and neurotransmitters originate in this area of the brain. It is also home to the appetite control center. Persons born with PWS face challenges in body functions, which are directly or indirectly impacted by hormones and neurotransmitters. There continues to be much more to learn about this area of the brain and its manifestations in PWS.

One of the most outstanding features seen in these individuals is their inability to control their food drive. The message of fullness never reaches their brain - they are always hungry. A chart has been included that will help you gain a better understanding of some of the areas affected by abnormalities in the hypothalamus in persons with PWS. The manifestation of characteristics and abnormalities can vary greatly among these students.



The Hypothalamus and Prader-Willi Syndrome

Altered Functions in Hypothalamus Seen in Persons with PWS	Explanations and/or Manifestations of Abnormalities
<p>ALTERED PITUITARY GLAND FUNCTION</p> <ul style="list-style-type: none"> • Decreased Growth Hormone • Altered Reproductive Hormones 	<p>This accounts for short stature, poor muscle tone, low metabolism and higher risk of osteoporosis. Growth hormone also helps to regulate blood lipids – putting people with PWS at higher risk for cardiovascular disease. A person with PWS can gain weight on half the calories of another person their age. They do not have the normal amount of muscle mass, which helps people burn calories. It often takes a diet of 600-800 calories to lose weight. Many children and young adults are now receiving growth hormone therapy so many of these abnormalities are being prevented or minimized.</p> <p>Very few people with PWS produce normal or even near normal levels of reproductive hormones. These young adults often start puberty but do not complete it. Women may never menstruate, and if they do, it is often irregular. Men may start to have voice changes and never outgrow this. Beard growth may be sparse. They often have an early onset of pubic and underarm hair growth. Hormone replacement therapy is often used. There have been a few – very rare cases where women with PWS have conceived and given birth to a child.</p>
DISRUPTION IN APPETITE CONTROL	<p>The hypothalamus houses the appetite control center. Depending on the individual, the drive for food can be mild to very severe. Typically individuals must rely on external supports to help them control this. Such supports must be present in all environments where food is present.</p>
ABNORMAL EMOTIONAL EXPRESSION	<p>The hypothalamus is one part of a system that helps with expression of anger and rage. Persons with PWS may have extreme fluctuations in emotions especially in the area of anger. This too is very individualized.</p>
<p>ALTERED REGULATION OF THE AUTONOMIC NERVOUS SYSTEM</p> <ul style="list-style-type: none"> • Temperature Regulation • Water Balance Regulation • Sleep Pattern Regulation 	<p>The hypothalamus controls our “thermostat” and other biological rhythms. Body temperature may elevate very quickly due to illness or high environmental temperatures. There are also cases where fever is absent in serious illness or infection. The hypothalamus also monitors and regulates body fluids by production of ADH (antidiurectic hormone). Researchers have documented abnormal sleep patterns in persons with PWS. This may contribute to daytime sleepiness.</p>
POOR AUDITORY SHORT TERM MEMORY	<p>The hypothalamus has input into our memory process. Persons with PWS often experience difficulty with short-term memory. One of their strengths however is their long-term memory. Once something is learned and stored in the long-term memory – it can be retrieved for years.</p>