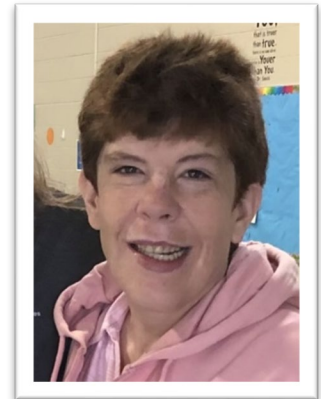


## Prader-Willi Syndrome (PWS) – FACT SHEET

### Adults with PWS

#### **Brief Overview of PWS**

- PWS is a very complex, neurobehavioral, genetic disorder caused by an abnormality on the 15<sup>th</sup> chromosome. Most of the challenges they face result from a defect in the area of the brain called the hypothalamus.
- It is a “spectrum disorder” meaning there are personal differences in the degree of challenges that these individuals face. They are individuals. They are not all the same.
- Due to advances in research and hormone replacement therapy, many of the younger adults with PWS may look different and face less challenges than older adults.
- It affects both males and females equally and in all races around the world.
- The prevalence range is 1:15,000 to 1:25,000 individuals.
- There are other genetic conditions or health issues that impact the hypothalamus which may require the same support as a person with PWS.
- There is no cure however, ongoing research continues. We are gaining more knowledge and understanding to help prevent and manage many of these challenges.



#### **The Hypothalamus and PWS**

PWS alters the function of the hypothalamus which impacts several major areas that can negatively impact the quality of life for these individuals.

- The appetite control center does not work properly. These individuals experience an uncontrollable appetite – the message of fullness never reaches the brain.
- There is also lowered hormone production including growth hormone and male /female reproductive hormones which result in poor muscle tone, short stature, low bone density and a lower metabolism. Other hormone deficiencies may be found.
- The hypothalamus helps to control emotions and behavior. It alters the “Fight or Flight” response in the autonomic nervous system. These individuals may experience an exaggerated response to stress, and it can cause a lack of impulse control.
- The temperature control center may be impacted. Many people experience a lower body temperature and may have other altered body temperature responses – both high and low.

#### **Common Features You May See**

- Poor muscle tone
- Short stature if not treated with growth hormone
- Learning and/or cognitive delays
- Incomplete sexual development
- Skin picking is common – may see open sores

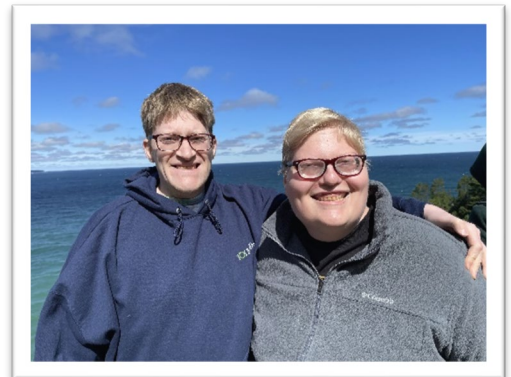
- Poor emotional control, obsessive compulsive tendencies, and other exaggerated stress behavioral responses. They are very impulsive and do not always understand consequences.
- Some have been found stealing – not just food items. (This may be due to a lack of impulse control.)

### **Health Concerns**

- High pain tolerance – they do not feel pain easily. Injuries and illnesses must be closely assessed and monitored.
- Gastrointestinal problems – constipation and slow stomach emptying common
- Swallowing difficulties – increased risk for choking
- Respiratory issues – hypoventilation (shallow, slow breathing), sleep disordered breathing, and sleep apnea are common.
- Sleep disturbances - daytime sleepiness, narcolepsy and cataplexy may be seen. Consultation with a sleep specialist is recommended.
- Orthopedic issues – higher incidence of hip dysplasia, scoliosis, osteoporosis
- Sensitivity to medications – especially those that cause sedation, including anesthesia, as well as those that slow the intestines (medicines used to treat diarrhea).
- Temperature control problems – may not run fever when infection is present, sensitive to high or low external temperatures
- Some may have mental health diagnoses – depression, OCD, Bi-polar disorder. Medications have been helpful in lowering and managing many of the manifestations of these disorders.

### **Supporting the Adult**

- Lifelong food security including constant supervision and planning for all sources of food; locks and alarms in most cases. 24-hour care and supervision are required.
- Daily exercise – helps to increase muscle mass, burn calories, and strengthen bones
- Low calorie diet to prevent weight gain
- Thrive on structure and routine. Prepare for changes. Other behavior support strategies may be needed.
- Some are successful in work and volunteer situations where food security and minimal changes are supported. Many are successful in a supported workplace where food is secure.
- Many adults are aging prematurely.
- As they age, we often see advancement in spine deformities. Many need walkers and other measures to prevent falls.
- They are typically very social individuals who love to go to the movies, take walks, attend dances, visit with friends, go shopping and play games.



We continue to learn more about the health and wellness needs of adults with PWS. Ongoing research continues as we strive to find a cure for the insatiable appetite as well as other challenges. People with PWS are living longer and have an improved quality of life. We are making progress.

For more detailed information about PWS, contact PWSA I USA at the email [info@pwsausa.org](mailto:info@pwsausa.org), the website [www.pwsausa.org](http://www.pwsausa.org), or phone number (941) 312-0400.