

Prader-Willi Syndrome (PWS) – FACT SHEET

Infants with PWS

Brief Overview of PWS

- PWS is a very complex, neurobehavioral, genetic disorder caused by an abnormality on the 15th chromosome. Most of the challenges these infants face result from a defect in the area of the brain called the hypothalamus.
- 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.
- It is a “spectrum disorder” meaning there are personal differences in the degree of challenges that these infants face. They are individuals. They are not all the same.
- Due to advances in research and hormone replacement therapy, many infants have experienced significant health and developmental benefits from growth hormone.
- There is no cure however, ongoing research continues. We are gaining more knowledge and understanding to help prevent and manage many of these challenges. There is hope for a cure.



Common Features You May See

- Infants with PWS are beautiful, happy babies.
- All experience poor muscle tone which often results in delayed developmental milestones. They may be slower to hold up their heads, roll over, crawl, and walk. However, you will see progress as their muscles grow stronger.
- They have weak muscles in their face and mouth, so they have a poor suck resulting in the need for specialized feeding techniques. Some may have a temporary feeding tube.
- Feeding and introducing solid foods takes time and patience. Nutrition is very important but, because of their muscle weakness, it often takes them time to master eating. It is important to provide a high fat diet. A nutritious high fat diet is important for all infants to assist in brain development.
- They tend to sleep a lot. They rarely cry and when they do, it may be weak.

Health Concerns

- Growth Hormone (GH) deficiency. There are numerous benefits for an infant to begin GH as soon as they complete an evaluation by a pediatric endocrinologist.
- It can be challenging for the infant to gain weight. Your primary care provider may have your baby come in for more frequent visits to make sure they are gaining weight and growing.
- Infant boys may experience undescended testicles. Discuss with the health care provider. A referral to a pediatric surgeon or urologist is recommended.
- Temperature regulation problems. They may not run fever when infection is present, and they are sensitive to high or low external temperatures.
- Respiratory issues – hypoventilation (shallow, slow breathing), sleep disordered breathing, and sleep apnea may be seen in some infants.



- Orthopedic issues – higher incidence of hip dysplasia and scoliosis. Consultation with a pediatric orthopedic specialist is recommended.
- Some experience eye muscle weakness resulting in cross eyes. Consultation with a pediatric ophthalmologist is suggested.
- Sensitivity to medications – especially those that cause sedation, including anesthesia.

Supporting the Infant

- Infants with PWS benefit from early intervention services to help prevent or slow developmental delays. It is common for them to receive therapy from an occupational, physical and speech therapist.
 - Because they sleep so much, parents must wake them at intervals and provide them with stimulation – singing, reading, talking, and moving around.
- Babies do not communicate their needs by crying so parents must check their diaper and schedule their feedings. Most set a timer.
 - Because of feeding and swallowing difficulties, many use specialized feeding techniques. There are special bottles and nipples that help them suck easier. Parents often must set a clock to know when it is time for a feeding. This will improve as their muscles get stronger.
 - Take time to enjoy your baby. Play and talk to them. Take them for walks. Celebrate every accomplishment.

You are never alone. Whether you are a parent, grandparent, friend, or other family member, PWSA I USA is here to help. Contact PWSA I USA to speak to a family support counselor or parent mentor who knows exactly what you are going through and who can answer your questions.

We continue to learn more about the health and wellness needs of infants with PWS. Ongoing research continues as we strive to find a cure for these challenges.

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