

Prader-Willi Syndrome *At a Glance*

(Pronounced Prah-der)

Prader-Willi syndrome (PWS) is a non-inherited genetic condition that happens as the result of a spontaneous mutation at the time of conception. There is nothing parents do that causes it and no practical way to prevent it.

Symptoms occur across a spectrum, with some individuals being more affected than others. Primary symptoms include an overwhelming desire to over-eat and a substantial risk for the development of obesity that starts in childhood. People with PWS often have cognitive, motor and language delays. There are distinct behaviors commonly associated with PWS. They can include temper tantrums, stubbornness, manipulation, and obsessive compulsive behaviors. People with PWS vary greatly in medical complexity, intelligence, and behaviors.

Although the cause is complex, PWS results from an abnormality on the 15th chromosome. It occurs in males and females equally and in all races. Prevalence estimates have ranged from 1:8,000 to 1:25,000 with the most likely figure being 1:15,000.

Common features that may be associated with PWS (not all people with PWS will have all of these features) include:

Low muscle tone

- Infants with PWS have low muscle tone. This generally improves over time.
- Adults often have decreased muscle bulk and tone.

Facial features

People with PWS have characteristic facial features.

- Narrow shaped head
- Almond shaped eyes
- Down turned mouth

Type 2 Diabetes

- Occurs in about 25% of adults with PWS.
- This may be a particular issue for those who are overweight.

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- The average onset of this type of diabetes is about 20 years of age.

Eye problems

- Could include strabismus (60-70%) or abnormal alignment of the eyes, squinting, and nearsightedness

Scoliosis

- Occurs in about 40-80% of people who have PWS.
- Varies in terms of age of onset and severity.

Respiratory problems

- Respiratory problems occur in about 50% of people with PWS, most commonly related to weak chest muscles that can lead to increased infections.

Bone fractures

- Increased rates of bone fractures due to reduced bone mass can occur.
- May also have leg edema and ulceration (especially in obese people).

Osteoporosis

- Osteoporosis can occur due to hormonal abnormalities and dietary limitations:
 - This often develops at a young age.
 - Walking and weight bearing exercise can be helpful in reducing the risk of bone density problems.
 - Consider calcium supplements to reduce the risk for osteoporosis.

Seizures in 10-20%

- Seizures occur in 10-20% of people with PWS

Oral health

- Dental cavities and other oral health problems may be increased.
- People with PWS often have thick saliva. This increases the risk of dental cavities.
- Good dental hygiene and regular check-ups are needed.

Decreased pain sensitivity

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- People with PWS have decreased pain sensitivity and a high pain threshold. This may mask signs of infection and/or injury.
- Many people will not complain, even with a significant infection or injury.

What you can do:

- Do not use report of pain as the only indicator of the seriousness of an injury or illness.

Bruising

- Many people with PWS bruise easily.
- It is important to note the cause and location of any bruises that occur during the day.

What you can do:

- Check all injuries. Document the injury location and communicate clearly with parents.
- Find out how the child handles pain. Some may exhibit increased fatigue and irritability which can serve as a clue to an underlying condition.

Abdominal illness

- Severe abdominal illness can occur as part of PWS.
- Abdominal bloating, pain, and vomiting may be signs of a life threatening stomach illness. If these symptoms are seen or reported, seek medical attention immediately.

What you can do:

- Monitor for bowel changes and problems.
- Work with parents and providers to ensure a high fiber diet with adequate intake of water.

Sensitivity to medications

- People with PWS may have increased sensitivity to medications.

What you can do:

- Consult with a doctor or other health care provider before giving medications that may cause sleepiness or drowsiness.

Skin picking

- Many children and adults exhibit skin picking.
- Open sores are common and can lead to infection.
- Insect bites can be the site of initial infection.
- Some children pick at various body openings.

What you can do:

- Provide diversion activities.
- Keep hands busy (coloring, computer etc.).
- Monitor frequent trips to the bathroom.
- Apply Band-Aids and teach basic first aid.
- Apply lotion (serves as a diversion and also moistures).
- Apply insect repellent when outside.

Appetite, anger, and temperature regulation

PWS impacts the functioning of the **hypothalamus**. This region of the brain is responsible for many important, automatic biological functions.

- The hypothalamus is involved in appetite control. People with PWS never feel full and are always hungry. The technical term for this is **hyperphagia** or an inability to control appetite. It is not something that a person with PWS can regulate themselves.
- The most challenging issues are overeating and obesity. Overeating usually begins between 1-6 years of age. Food seeking behaviors include: hoarding or foraging for food, eating inedible foods, and stealing food or money for food. Poisoning, and food poisoning in particular, as well as choking are significant risks.
- People with PWS have a reduced energy requirement. They need a low calorie, well balanced diet with strict supervision of food access. This needs to be combined with regularly scheduled exercise and activities.

What you can do about appetite:

- Work with a nutritionist to implement a calorie restricted diet.
- Supervise and monitor all food. Keep food and money out of sight (money buys food). Lock cupboards and refrigerators.
- Weekly weight monitoring by school nurse may be helpful.
- Growth hormone therapy may help.

- Monitor eating as they may eat quickly and choke.
- Provide careful supervision to ensure that people with PWS are not being taken advantage of by others in exchange for food.

The hypothalamus also helps control anger and rage. People with PWS often have abnormal emotional expressions and extreme bouts of anger.

It is also involved in the regulation of temperature, water balance, and sleep patterns.

It controls our body thermostat. Body temperature may elevate very quickly with illness and high temperatures (see next bullet).

- People with PWS often have altered temperature regulation. Therefore unexplained high and low body temps are common.
 - People may have little or no fever present, even with severe infections.
 - May have a quick response to high or low outside temperatures and exposure should be limited.
 - Make sure they do not get overheated or too cold.
 - If you suspect a child is ill, contact parents/nurse.

Decreased growth hormone

People with PWS have decreased levels of growth hormone.

- This accounts for short stature, poor muscle tone, low metabolism, and high risk of osteoporosis.
- Growth hormone also helps regulate blood lipids, increase bone density, increase muscle mass, and increase strength and endurance.
- Growth hormone may help with the regulation of breathing during sleep.
- Growth hormone replacement therapy is common among many people with PWS but should only be started after a careful medical evaluation by an experienced clinician.

Altered reproductive hormones

- Very few people with PWS produce normal or even near normal levels of reproductive hormones.
- They will often start puberty but not complete the process. They will often have early onset pubic and underarm hair growth.
- Girls may never menstruate or have irregular menstruations.
- Boys start to have voice changes but never start a growth spurt.

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- It is important to provide appropriate sex education.

Things to Think About

1. Medical / Dietary Needs

What you need to know

The list of *possible* medical problems in PWS can be quite extensive. However, each individual usually has only some of these problems. Also, the severity of any one of these medical problems varies widely between individuals. Therefore, it is important to ask the parents for information about the medical issues affecting their child.

School age children with PWS may have annual doctor and specialist visits to monitor medical conditions.

What you can do

- Encourage parents to get a yearly check- up for the child with appropriate screening, diagnosis and treatment of co-occurring conditions.
 - should be centered in the child's primary care Medical Home
- Ensure the child's vaccination history is up-to-date. Most children with PWS can receive live virus vaccinations. Recording information about the types of vaccinations the child receives is important.
- Support good hand washing to minimize the spread of viruses in the classroom.
- Notify the parents of any changes in the child's energy level.
- Be aware of any changes in behavior or mood that seem out of line with the situation. Notify the parents.
- It is important to be aware of any academic changes. Contact parents when any differences are noticed.
- Be an advocate for the child in obtaining sufficient speech and language support during school to communicate effectively throughout the day. This may include communication augmentation devices in some situations.
 - Work with parents when they are advocating for their child.
- Work with parents to establish behavioral and environmental support for weight management. Consider:
 - Restricted access to food in all environments
 - Locks on refrigerators and food storage areas (including other student's lunches and snacks)

- Constant supervision
- Calorie restrictive diets
- Consistent and scheduled meals and snacks
- Programs that help teach behavioral and weight management strategies.

2. Education Supports

It is important to have high learning expectations for children with Prader-Willi. Encourage use of the core educational curriculum and modify it in order to meet the individual needs of the child.

What you need to know

Appropriate supports

Many children do well in regular classrooms as long as they have the appropriate supports to be successful.

- The IEP team will help determine what supports and modifications are needed for the child with PWS to be successful.

Cognitive issues

Cognitive issues include:

- Global intellectual disability
- People with PWS typically have mild intellectual disabilities (IQ 60-70s), although higher IQs have been reported.
- Regardless of IQ, most children with PWS have multiple complex learning disabilities and difficulties with academic performance.

Relative Strengths

Many students with PWS show relative strengths in reading/decoding, expressive vocabulary, spatial-perceptual organization, long term memory, and visual processing.

- Not every child with PWS shows these patterns however, and teaching strategies should always be based on individualized test profiles of strengths and weaknesses.

- Even when test scores are in the normal range, most children cannot generalize from one experience to the next. This often becomes evident late in primary grades.

Overall cognitive profile

Overall cognitive profile includes: cognitive rigidity, attention deficits, and problems with short-term memory, auditory processing, sequential processing, arithmetic and social cognition.

- An individual may present with problems in reading, math, spatial ability, neatness, test taking, speech difficulties, and/or the ability to make friends and communicate effectively with peers.

Learning disabilities

Learning disabilities result in associated short-term memory and sequencing deficits.

- Sequencing and language difficulty underlies many behavioral changes. This may make it hard to recognize cause and effect. It may be hard to link rewards and consequences to behaviors. Environmental management is often more effective than behavioral management. Consider positive behavioral supports.
- Difficulty applying knowledge in new situations may prevent some children from using facts in a practical or productive way.

Communication

- People with PWS often have a specific motor speech disorder called apraxia of speech.
 - This leads to difficulty with the sequencing and coordination of motor speech movements necessary to articulate clear and concise speech.
 - Children with PWS may experience difficulty feeding in the early stages of life and experience, Failure to Thrive because of these difficulties.
- Speech articulation issues are often contributed to low muscle tone, poor motor planning, and thick saliva.
 - More delays occur with expressive language compared to receptive language.

What you can do

- Incorporate language skills in social, work, and life skill areas.
- People with speech and motor difficulties often benefit from speech and occupational therapy.
- Teach learning strategies for non-verbal communications.

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- Consider new technology, computers, sign language.
- People with PWS may struggle with writing, cutting, and other fine motor tasks.
- PT and OT can help students improve their strength and physical abilities.
- Identify physical activities that can be enjoyed and used throughout a student's life.
 - Examples: Biking, skating, jumping on trampoline, dance, and ball playing.

3. Behavior and Sensory Support

What you need to know

Behavioral and psychiatric problems

- Behavioral and psychiatric problems may interfere with quality of life for adolescents and in adulthood.
- These behaviors can be persistent. They may escalate in severity and intensity with age (particularly from adolescent to adulthood).
- This escalation is independent of intellectual, language, and motor abilities.

Characteristic behavior profile

- People with PWS have a characteristic behavior profile which includes: temper tantrums, stubbornness, emotional lability, exaggerated emotional response, controlling and manipulative behaviors, compulsivity, and difficulty with changes in routine.
- Many behavioral challenges are similar to those found in people diagnosed with autism or autism spectrum disorders.
- Attention Deficit Hyperactivity Disorder (ADHD) may be seen.
- Perseveration and repetitive behaviors are common.
- Students with PWS can be demanding and prone to rage episodes when frustrated.

Psychosis

- Psychosis is reported in young adulthood in 10-20% of people who have PWS.
- By late adolescence, about 15% will have a diagnosed mood disorder.
- Delusions and paranoid ideation are more common in people with PWS than in the general population.
- They may have obsessive compulsive tendencies.

- These behaviors can be a source of strain in families, and in living and work environments.

Monitoring food

- The constant need to monitor food intake can be very stressful for families.
- This is due to an abnormally increased appetite and the eating of unusual food related items (i.e. sticks of butter, used cooking grease, decaying food, garbage, and food flavored items like shampoo).
- Hoarding food when it is accessible is common.

Judgment about social cues

- Social judgment can be poor. Interpretations of social information by students with PWS are similar to some children who have a pervasive development delay.
- They may have an inability to read other's facial expressions, decode emotion, and have difficulty interpreting visually presented social information.

Sensory issues

- Decreased sensitivity to pain
- Tendency to not run a fever even when significantly ill
- Increased skin picking and picking at other body areas

What you can do

Behavioral support strategies

- Consider proactive behavioral support strategies with behavioral or mental health professionals
- Parents may consider a referral for medication for a child.
 - Selective serotonin reuptake inhibitors (e.g., fluoxetine and sertraline) have been helpful in many patients.
- Be firm with directions, rules, and discipline.

Weight management

- Behavioral and environmental support for weight management is needed.
- Consider:
 - Restricted access to food in all environments

- Locks on refrigerators and food storage areas (including other student’s lunches and snacks)
- Constant supervision
- Calorie restrictive diets
- Consistent and scheduled meals and snacks
- Programs that help teach behavioral and weight management strategies.

Regulating emotions and behavior

- Many children and young adults with PWS have difficulty regulating emotions and behavior. This is especially true when handling unplanned changes.
- They usually thrive with consistency and routine. They can be easily upset with disruption.
- Prepare students for any change in schedule.
- Provide a “safe” area for students to share emotions.
- Teach and model use of words and/or pictures in sharing emotions.
- Teach and emphasize behaviors you want.
- Make sure they have an effective communication system.

Skin picking

- Skin picking helps them decrease anxiety and boredom.
 - Eliminate opportunities to skin pick.
 - Distraction may be helpful.
 - Decrease anxiety and eliminate boredom.

Social cues and coaching

- Provide social cues and coaching.
- Provide information and discuss differences with the child’s peers.
- Help develop confidence and focus on strengths.
- Provide positive reinforcement.
- Teach child appropriate social behaviors/skills (how to ask a friend to play, role model, small groups of friends).
- Teach how to recognize facial expressions, body language, and moods.

4. Physical Activity, Trips, Events

What you need to know

- People with PWS will need to be monitored to avoid foraging for food or stealing money to buy food.
- During meal times, it is important to watch child's eating as choking has been reported
- Any change in routine may produce anxiety, fears, and/or worry. Even a fun activity can produce anxiety and resulting behavioral challenges. Crowds and loud noise may be overwhelming to some people.
- If you live in New England (USA) and qualify, Northeast Passage offers Therapeutic Recreation and Adaptive Sports programming (www.nepassage.org).

What you can do

- Be proactive and discuss any change in schedule or setting with the child ahead of time.
- Use social stories and pictures to help them understand the change.
- Role-play different social settings and appropriate behavior.

5. School Absences and Fatigue

What you need to know

- No excessive absences are expected.
- Sleep abnormalities are common in individuals with PWS.
 - These sleep abnormalities include: reduced REM (rapid eye movement), altered sleep architecture, oxygen desaturation and both central and obstruction apnea.
- Many students with PWS have daytime sleepiness.
 - This may be the result of altered breathing patterns in the hypothalamus, poor muscle tone, and weak chest muscles. Upper airway obstruction may also contribute to sleep abnormalities.
 - If people with PWS snore, sleep walk, or have persistent morning headaches, they should be evaluated for sleep apnea.
- Lack of sufficient sleep can lead to behavior issues and temper tantrums.

What you can do

- Focus on improving nighttime sleep.
- Schedule daytime naps or afternoon rests if needed.

- Some children respond to a change in scenery, i.e. taking a walk.

6. Emergency Planning

What you need to know

Emergency plans will need to be individually determined with the input of the parents, based on the child's specific behaviors and medical issues. It is important to mention any new signs, symptoms, and/or pain to the child's parents.

7. Resources

Gene Tests

www.ncbi.nlm.nih.gov/sites/GeneTests/

Learn more about the genetics of Prader-Willi syndrome. GeneTests is a publicly funded medical genetics information resource developed for physicians, other healthcare providers, and researchers, available at no cost to all interested persons.

Genetics Home Reference

<https://ghr.nlm.nih.gov/condition/prader-willi-syndrome>

Consumer-friendly information about human genetics from the US National Library of Medicine

Prader-Willi Syndrome Association (PWSA)

<http://www.pwsausa.org/>

Prader-Willi Syndrome Association (USA) is an organization of families and professionals working together to raise awareness, offer support, provide education and advocacy, and promote and fund research to enhance the quality of life of those affected by Prader-Willi syndrome.

A recommend resource available through the Prader-Willi Syndrome Foundation is:

The student with Prader-Willi Syndrome Information for Educators, by Barbara Dorn and Barbara J. Goff, Ed.D order through PWSA, <http://www.pwsausa.org/>

Foundation for PWS Research

<http://fpwr.org/about-prader-willi-syndrome>

The mission of Foundation for PWS Research is to eliminate the challenges of Prader-Willi syndrome through the advancement of research. High-quality research will lead to more effective treatments and an eventual cure for this disorder. By working together, we intend to free our loved ones from the burdens of PWS, allowing them to lead full and independent lives.

Classroom Accommodations for Students with Visual Issues

Classroom Accommodations for Students with Visual Issues is a resource from Boulder Valley Vision Therapy, P.C. (<https://www.bouldervt.com/>), in Boulder, Colorado. It describes a variety of variety of issues and offers strategies.

<https://www.bouldervt.com/wp-content/uploads/sites/478/2015/12/227-Course-Handout-CCIRA-2016.pdf>

Note: *This printable version does not include the information found under the green button marked "Transitions" on the website. Those general pages may be printed separately.*

8. Meet a Child with PWS

Meet Emma!

GEMSS would like to thank Emma and her mother for their generosity in sharing this story with us. You have made the site come to life with the addition of your thoughts and feelings. Thank you so much!

Emma is an enthusiastic 11 year old who loves to ask questions. As a 6th grade student in rural New Hampshire (USA), she is in regular classes. She has two older brothers and a sister. She is very comfortable in her home environment and loves to play with her pet dogs and her dolls. "That is when she is happiest," says



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her mother, Lynn. She likes listening to hip-hop and country music and is learning to play the clarinet. She learned how to ride a bike this year! Her older sister helps provide Respite to the family. She gets along well with the customers in her mother's beauty salon and engages them in conversations.

Emma was diagnosed with Prader-Willi Syndrome when she was 18 months of age.

Emma has supports to help her keep on task and learning during school. Part of keeping her safe in school is making sure she can't access other people's snacks and lunches, as "food-seeking" is a daily struggle. This has to be explained in detail in new school situations, Lynn feels. For example, telling the school-based team that she is a food-seeker is not enough. Details such as 'she will go into the garbage or even dumpsters to seek food' really helps to drive home the importance of safety monitoring at all times. Another example of explaining the food-seeking behavior in detail is that "Emma will have a hard time focusing in class if there is popcorn popping in another classroom".



One day, Lynn read some negative comments in the communication book that travels between home and school, saying Emma was "defiant, not focused, and not paying attention." Lynn did some great detective work by finding out what was happening in the school. She learned that snacks were taken out of all the children's backpacks to keep them from Emma. However, they were still contained in a corner of the classroom, and even though Emma didn't seem like she saw those hidden snacks, she did know exactly where the food was and couldn't concentrate on her work with the competing food in the near environment. So, by changing the

environment (locating the food in a distant classroom), the team was able to eliminate the 'behaviors' and the next day, all was well with Emma.

Academically, Emma is most challenged with math, social studies, and science, as some of the concepts are hard for her to grasp. Comprehension is an issue but her team helps her to learn how to problem solve. For example, if Emma has to know the population of a state, they might help her learn how to search for the answer by looking for a sentence that has the word 'population' in it. On field trips, Lynn says the other students help her to understand the tasks



and they seek help if they are having a hard time conveying an idea. Emma is happy with her social relationships and says, that “everyone is her friend.”

Medically, a challenge in school has been a ‘narcolepsy-like’ disorder, which made Emma fall asleep in class. Now she is on a medication to help her stay alert and awake but they had to do some problem solving to figure out this issue.

Lynn has some advice for other parents and teachers and suggests to all that you, “expect the unexpected” as bumps in the road will happen. She says it is hard to find people who feel comfortable with Emma but she uses her resources and her older daughter has been exceptional in helping out. Lynn says that teams need to be patient and work together to make sure that the children are safe and comfortable in school. Using resources such as outside experts to help the team is fine with Lynn – whatever it takes to make sure people really understand her daughter’s needs.