

## Prader-Willi Syndrome (PWS) *For Healthcare Providers*

This is a customized health care provider version of our website. Please visit the main website to find more comprehensive information for families and schools ([www.gemssforschools.org](http://www.gemssforschools.org)).

### Physical characteristics and/or symptoms

*Note: not all people with Prader-Willi syndrome will have all of these features.*

- Low muscle tone
- Facial features
  - Narrow shaped upper part of the head
  - Almond shaped eyes
  - Down turned mouth
- Type 2 Diabetes
- Eye abnormalities
  - Strabismus
  - Abnormal alignment of the eyes
  - Nearsightedness
- Scoliosis
- Respiratory problems
- Bone fractures
- Osteoporosis
- Seizures
- Oral health
  - Dental cavities and other oral health problems may be increased.
  - People with PWS often have thick saliva increasing the risk of dental cavities.
- Decreased pain sensitivity
- Bruising
- Abdominal illness
- Sensitivity to medications
- Skin picking
- Appetite, anger, and temperature regulation
  - PWS impacts the functioning of the *hypothalamus*.

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- People with PWS never feel full and are always hungry. This hyperphagia is not something that a person with PWS can regulate themselves.
- The most challenging issues are overeating and obesity.
- 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.
- People with PWS often have abnormal emotional expressions and extreme bouts of anger.
- People with PWS often have altered temperature regulation.
- Decreased growth hormone
- Altered reproductive hormones
  - Puberty is started but not completed. 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.

## Recommended Routine Surveillance

- Strict supervision of daily food intake and exercise program
- Annual testing for hypothyroidism
- Ophthalmologic evaluation
- Management of hypotonia
- Evaluation of respiratory and sleep status
- Scoliosis evaluation
- Monitor for seizures
- Management of skin picking behavior

## Emergency Protocols

There are no specific emergency protocols for this particular condition as it is not typically associated with episodes of sudden and serious medical decompensation.

- Emergencies should be handled as with any child.
- If seizures are present, the following seizure action plan may be useful:

[https://www.aap.org/en-us/Documents/Seizure Action Plan for%20School.pdf](https://www.aap.org/en-us/Documents/Seizure_Action_Plan_for%20School.pdf)

## Specialists Who May Be Involved

Follow up is need on a case-by-case basis. A multidisciplinary team approach to best meet the child's individual needs is recommended.

- Behavior and psychiatric evaluation
- Dentist
  - Dental anomalies
- Developmental evaluation:
  - Speech therapy
  - Physical therapy
  - Occupational therapy
- Endocrinologist
  - Hypothyroidism
  - Hypogonadism
  - Growth hormone treatment
- Gastroenterologist
  - Feeding problems
- Geneticist/genetic counselor
  - Diagnosis
  - Coordination of care
  - Genetic risk for family
  - Clinical trials
- Neurology
  - Monitor seizures
- Nutritionist
  - Management of diet and exercise program
- Orthopedists
  - Hip dysplasia
  - Scoliosis
  - Increased risks for bone fractures due to osteopenia and osteoporosis
- Ophthalmologist
  - Strabismus
- Sleep specialist
  - Sleep issues

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## Sample Forms

- Sample paragraph to be used for Letters of Medical Necessity or Letters to the school:

*My patient \_\_\_\_\_ has been diagnosed with Prader-Willi syndrome (PWS). This condition is characterized by excessive eating, global developmental delays and behavioral abnormalities. Medical complications with PWS include management of weight gain and behaviors. Because of these, \_\_\_\_\_ needs the following accommodations.*

## Seven Important Aspects of School Life

“[Prader-Willi Syndrome at a Glance](#)” will help you talk with parents and schools about:

- Medical / Dietary Needs
- Education Supports
- Behavior & Sensory Supports
- Physical Activity, Trips, Events
- School Absences & Fatigue
- Emergency Planning
- Transitions



## Resources

### Genetic Home Reference

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

### GeneReviews

<https://www.ncbi.nlm.nih.gov/books/NBK1330/>

### AAP Health Supervision for Children with Prader Willi Syndrome

<http://pediatrics.aappublications.org/content/pediatrics/127/1/195.full.pdf>

**Additional resources including support group information can be found on the main website.**