

# Prader-Willi Syndrome: What You Need to Know

In this handout, you will learn about Prader-Willi syndrome and its symptoms. You will also learn how doctors diagnose and treat Prader-Willi syndrome.

## WHAT IS PRADER-WILLI SYNDROME?

Prader-Willi syndrome (PWS) is a rare genetic condition that affects your child's appetite, growth, **metabolism** (how their body uses food for energy), and behavior. Babies with PWS are very weak and have trouble feeding. Later in infancy and during childhood, they often do not feel full after a meal. This can cause unhealthy weight gain and behavior challenges.

## WHAT CAUSES PWS?

PWS is caused in several ways:

- When a small amount of genetic material on chromosome 15 is not inherited from the father
- A missing piece of chromosome 15
- A gene that is changed on chromosome 15.

In most cases, PWS happens at random. **It is not anyone's fault.**

These genetic changes usually lead to changes in the **hypothalamus** (part of the brain that controls the release of hormones). When the hypothalamus does not work properly, it can affect your child's appetite, growth, development, mood, and sleep.

## WHAT ARE THE SYMPTOMS OF PWS?

Symptoms of PWS are different in every person. Not everyone is affected the same way. Symptoms can also change with age.

### Babies

- Low muscle tone (hypotonia)
- Difficulty sucking with trouble feeding
- Distinct facial features, such as a narrow nasal bridge; almond-shaped eyes; narrow head; a turned-down mouth; and a thin upper lip
- Delayed or poor development of motor skills, such as feeding, sitting up or walking

### Childhood, teen years and adulthood

- Constant food cravings and a slow metabolism. The cravings and slow metabolism can cause unhealthy weight gain or obesity if left unchecked.
- Learning disabilities
- Problems or delays in learning language
- Behavioral issues
- Little or no sex hormone production from the brain. This can cause small sex organs, incomplete or delayed puberty, and infertility (trouble reproducing later in life)
- Short stature (short height)
- Small hands and feet
- Scoliosis (curved spine)

## HOW DO DOCTORS DIAGNOSE PRADER-WILLI SYNDROME?

- Review of your child's medical history
- Physical exam
- Genetic testing

## HOW DO DOCTORS TREAT PWS?

There is no cure for PWS. However, doctors can often treat some of the conditions that go along with PWS. The doctor will talk with you about creating a treatment plan for your child's needs and symptoms.

Common PSW treatments include:

### Therapies and medications

- **Various types of therapy.** This can include speech therapy, occupational therapy, and physical therapy. These therapies can help with your child's development.
- **Behavior management.** Children with PWS can develop challenging behavior issues. A psychologist can be helpful in finding your child the support they need.
- **Weight management.** A dietitian can help you make a healthy diet plan to help manage your child's weight and make sure they are getting the nutrition needed.
- **Human growth hormone (HGH) treatment.** This might help your child grow taller and have healthy levels of body fat. It can also help with movement and breathing. An endocrinologist (hormone doctor) can help discuss this option with you.

### Follow-up care and check-ups

- **Yearly eye check.**
- **Checking your baby's nutrition and feeding skills.**
- **Scoliosis check.** Every year, your child should be evaluated for scoliosis (a curve in the spine) by their primary care physician.

### Tests

- **Bloodwork.** Every year, your child's doctor should check the blood for thyroid levels.
- **Sleep study.** If your child has trouble sleeping, an overnight sleep study in the hospital can be helpful.

### Surgery

- **Orchidoplexy surgery.** In boys with PWS, sometimes the testicles do not fully descend (drop down into the proper position). In such cases, a small surgery can bring them down to their proper location.

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## Medical Genetics

Mass General Hospital for Children  
55 Fruit Street, Suite 6C  
Boston, MA 02114

[www.massgeneralforchildren.org/genetics](http://www.massgeneralforchildren.org/genetics)

For more information please call

617-726-1561