CONDITION

Prader-willi syndrome

Prader-willi syndrome is a lifelong genetic condition affecting appetite, growth, development, hormones, and behaviour.

WHAT IT IS
Lifelong genetic condition affecting development, appetite, growth, and hormones

BODY SYSTEM
Brain, hormones, muscles, growth, and metabolism

MAIN SYMPTOMS
Weak muscles, constant hunger, learning difficulties, short stature

WHAT CAUSES IT
Loss of paternal chromosome 15 gene activity

HOW IT IS DIAGNOSED
DNA methylation testing, followed by subtype testing

MAIN TREATMENT
Multidisciplinary care, food security, hormone treatment, developmental support

CAN IT BE CURED?
No - genetic changes cannot be reversed

IS IT AN EMERGENCY?
Sometimes - choking, breathing difficulty, or severe abdominal symptoms

LONG-TERM OUTLOOK
Lifelong support with many preventable complications

Overview

Prader-willi syndrome, usually written as Prader-Willi syndrome or PWS, is a lifelong genetic condition. It affects how the brain controls appetite, growth, muscle tone, hormones, sleep, emotions, and behaviour.

Babies with PWS often have very weak muscles, called hypotonia. They may be unusually sleepy and struggle to suck or gain weight. During childhood, appetite usually increases. A person may rarely feel full, even after eating enough. This is called hyperphagia.

Without a structured food environment, hyperphagia can lead to rapid weight gain and serious health problems. It is not caused by poor self-control or parenting. The brain does not send or process fullness signals in the usual way.

PWS also affects learning, physical development, and the production of sex and growth hormones. The pattern and severity are different for each person. Early diagnosis allows feeding, development, nutrition, sleep, and hormone needs to be addressed promptly.

Symptoms

Signs of PWS change with age.

During pregnancy, there may be reduced fetal movement. Some babies are born in a breech position. After birth, common features include:

Feeding problems may require specialist support. Some babies need tube feeding for a period, although feeding ability usually improves.

In early childhood, interest in food begins to increase. Over time, a child may constantly think about food, seek it out, eat very quickly, or become distressed when access is limited. Weight gain can occur on fewer calories than expected because PWS often reduces muscle mass and energy use.

Other possible features include short stature, small hands and feet, reduced strength, and a narrow forehead. Puberty may be late or incomplete because the ovaries or testicles produce too little sex hormone. Boys may have undescended testicles. Fertility is usually reduced, although pregnancy is possible in some women.

Learning ability ranges widely. Many people have mild to moderate intellectual disability, while others have average intelligence with specific learning difficulties. Speech, movement, planning, and flexible thinking may be affected.

Common behavioural or emotional concerns include anxiety, repetitive questions, difficulty coping with unexpected change, skin picking, and intense distress around food. Some people have features of autism or attention-deficit hyperactivity disorder. Mood or psychotic illnesses can occur, particularly in some genetic subtypes.

Sleep apnoea is also common. This means breathing repeatedly pauses or becomes shallow during sleep. Signs include loud snoring, restless sleep, daytime sleepiness, morning headaches, or pauses in breathing noticed by someone else.

- Very low muscle tone and a weak cry
- Difficulty sucking, swallowing, or gaining weight
- Unusual sleepiness and reduced movement
- Delayed sitting, standing, walking, or talking

Causes and risk factors

PWS occurs when a group of genes on chromosome 15 does not work as expected. Chromosomes are packages of genetic information inside cells.

For these particular genes, the working copies normally come from the father. In PWS, that paternal gene activity is missing. This may happen because:

Genomic imprinting is the natural process that switches certain genes on or off according to which parent they came from.

Most cases happen by chance and are not caused by anything either parent did before or during pregnancy. PWS affects people of all sexes and ethnic backgrounds.

The chance of PWS occurring again in a family is usually low, but it is higher with certain uncommon imprinting or chromosome changes. Genetic counselling can explain the specific result and what it may mean for parents, siblings, or future pregnancies.

- Part of the paternal chromosome 15 is deleted
- Both chromosome 15 copies came from the mother
- An imprinting problem prevents the paternal genes from switching on
- Rarely, a chromosome rearrangement affects the region

Diagnosis

Symptoms may suggest PWS, but genetic testing is needed to confirm it.

The usual first test examines DNA methylation in the PWS region of chromosome 15. Methylation is a chemical marker that helps control whether genes are switched on. This test can identify the abnormal imprinting pattern found in almost everyone with PWS.

Further genetic tests are needed to find the subtype, such as a deletion, two maternal copies of chromosome 15, or an imprinting defect. Knowing the subtype can clarify the chance of recurrence and may help the care team anticipate some health or behavioural needs.

After diagnosis, assessments commonly cover feeding and swallowing, growth, nutrition, development, hearing, vision, hormone function, sleep, and the spine. These are health checks for known features of PWS; they do not mean every complication will occur.

Treatment and management

There is no treatment that reverses the genetic change. Management focuses on development, physical health, quality of life, and preventing complications.

In infancy, feeding support may include positioning, specialist bottles, swallowing assessment, and temporary tube feeding. Calorie intake needs careful review because feeding difficulty can change into excess weight gain as the child grows.

A planned, secure food environment becomes central to care. This often means predictable meals, clear rules shared by all caregivers, supervised access to food, and limiting unscheduled eating. Some families need locked food storage. These measures protect the person from a biological drive to eat; they are not punishment.

Regular physical activity supports strength, mobility, bone health, sleep, and weight management. Physiotherapy and occupational therapy can help with low muscle tone and daily skills.

Growth hormone treatment is commonly considered for children and may be considered for some adults. It can improve growth, body composition, muscle strength, and physical function. Assessment and monitoring are needed, including attention to breathing during sleep, blood sugar, growth, and the spine.

Other hormone treatments may be offered for low thyroid hormone, reduced sex hormones, or other confirmed deficiencies. Treatment is based on test results, age, development, and individual risks.

Speech and language therapy, educational support, behavioural strategies, and mental health care may also be useful. Predictable routines and advance warning of changes often reduce anxiety. Skin picking may improve with wound protection, keeping hands occupied, and behavioural support.

Care is usually coordinated across genetics, hormone care, nutrition, sleep medicine, development, mental health, and primary care.

Complications

Many complications are linked to hyperphagia, reduced muscle mass, hormone differences, or sleep problems. They are not inevitable.

Excess weight can contribute to type 2 diabetes, high blood pressure, fatty liver disease, joint problems, heart disease, and breathing difficulties. Sleep apnoea can occur at any body size and may worsen daytime behaviour, concentration, and heart health.

Scoliosis, a sideways curve of the spine, may develop or progress during growth. Low sex

hormones and reduced activity can weaken bones, increasing the risk of osteoporosis and fractures.

Choking is a concern because some people eat rapidly, take large mouthfuls, or have swallowing difficulties. Reduced vomiting and an unusually high pain threshold may make abdominal illness harder to recognise. Sudden abdominal swelling, pain, vomiting, marked tiredness, or refusal to eat can occasionally signal a serious stomach or bowel problem.

Skin picking can cause wounds, scarring, or infection. Mental health complications may include severe anxiety, depression, or psychosis. A sudden major change in behaviour, sleep, thinking, or functioning deserves medical assessment rather than being assumed to be part of PWS.

Prevention

PWS usually cannot be prevented. It results from a genetic event that is generally outside anyone's control. Food choices, medicines, stress, or actions during pregnancy do not cause it.

Genetic counselling cannot prevent PWS, but it can explain the genetic subtype and the chance of it happening in another pregnancy. Prenatal or pre-implantation testing may be available in some situations, particularly when a family has a known chromosome or imprinting change.

Complications can often be reduced through early diagnosis, secure access to food, appropriate nutrition, physical activity, sleep assessment, hormone care, and regular health monitoring.

Living with it and what to expect

PWS changes over time. Severe feeding difficulty and low muscle tone dominate infancy. Appetite and food-seeking usually become more prominent during childhood and remain lifelong. A person does not generally grow out of hyperphagia, even if their weight stays within a healthy range.

Development continues throughout childhood and adulthood, although skills may be learned more slowly. With suitable teaching and therapies, many people improve their communication, movement, self-care, and social skills.

Predictability is often important. Consistent meal routines, clear expectations, visual schedules, and calm preparation for changes can reduce stress. Plans work best when home, school, respite care, and community services follow the same food-safety approach.

Most people need lifelong help managing food access, healthcare, money, transport, or decision-making. Some adults can work, volunteer, form close relationships, and take part in community activities. Independent living without food supervision is uncommon, but supported housing can provide both safety and meaningful independence.

Long-term health is strongly influenced by weight, breathing during sleep, diabetes risk, mobility, and access to coordinated care. Serious complications can shorten life, but they are not unavoidable. Ongoing follow-up aims to identify changes early while supporting education, relationships, activity, and personal goals.

When to get medical help

Arrange routine medical follow-up for growth, weight, nutrition, development, hormone function, sleep, behaviour, bone health, and scoliosis. Seek a timely appointment if there is new snoring, daytime sleepiness, rapid weight change, reduced mobility, worsening skin wounds, or a lasting change in mood or behaviour.

Get urgent medical advice for:

Call emergency services for choking, severe breathing difficulty, blue or grey lips, collapse, a seizure lasting several minutes, or inability to wake the person.

People with PWS may not show pain, fever, or vomiting in the usual way. A clear change from their normal behaviour or energy level can therefore be medically important.

- New abdominal pain, swelling, vomiting, or refusal to eat
- Fever with unusual sleepiness or reduced responsiveness
- A skin wound that becomes increasingly red, swollen, painful, or draining
- A sudden major change in behaviour, thinking, or sleep
- Symptoms of very high blood sugar, such as excessive thirst and urination

Common questions

Is Prader-Willi syndrome inherited?

Usually, no. Most cases result from a new genetic event in the egg, sperm, or early embryo. Some rare imprinting or chromosome changes can run in families. Subtype testing and genetic counselling provide the most accurate family-specific information.

Can someone with PWS ever feel full?

Many people have weak or absent fullness signals and remain preoccupied with food after eating enough. The intensity differs, but hyperphagia is usually lifelong. A secure food environment is often needed even when weight is well managed.

Can a person with PWS live independently?

Some people develop strong self-care, work, and social skills, but most need continuing support. Unsupervised access to food can remain unsafe. Supported living can offer choice and independence while maintaining food security and healthcare oversight.

What is the life expectancy for PWS?

There is no single figure that applies to everyone. Obesity, breathing problems, diabetes, choking, and serious abdominal complications can shorten life. Early diagnosis and consistent management of weight, sleep, hormones, and other health needs can improve long-term health.

Why do babies with PWS struggle to feed but later overeat?

The condition affects brain systems that control muscle tone, feeding, appetite, and energy use. Weak sucking and sleepiness cause early feeding difficulty. As the brain's appetite regulation changes, hunger and food-seeking become more prominent.

Is there a cure for PWS?

There is currently no cure that restores the missing gene activity. Treatments can still make a meaningful difference by supporting growth, strength, development, sleep, hormone health, behaviour, and safer management of appetite.

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