



Empowering Children. Supporting Families. Building Brighter Futures.

Resource 068 | Genetic and chromosomal conditions

Prader-Willi syndrome

What it means

Prader-Willi syndrome is a genetic condition with changing needs across childhood. Infants often have low muscle tone and feeding difficulty. Later, an unusually strong drive to eat can develop, alongside growth, hormonal, learning, and behavioral concerns. Support requires both medical care and a consistent, respectful environment. [1]

Causes and risk factors

The condition results from loss of expression of particular genes normally active on the paternally inherited chromosome 15. Several genetic mechanisms can cause this. Appetite and hormonal differences are biological features, not a failure of willpower. Genetic counseling explains the specific mechanism and family implications. [1,2]

General family education. A concern deserves assessment, but this guide does not diagnose a child or prescribe treatment.



Signs and everyday impact

Infants may be very floppy, sleepy, and unable to feed effectively. Later features can include delayed milestones, short stature, increased appetite, food seeking, sleep problems, and learning or behavioral difficulties. Nutritional needs change with age; an infant's feeding problem should not be managed like later hyperphagia. [1,2]

Assessment and diagnosis

DNA methylation testing and additional genetic tests establish the diagnosis and mechanism. Assessment covers growth, nutrition, endocrine function, breathing during sleep, development, and behavior. Regular review is important because needs change. A dietitian and endocrinologist help interpret growth and plan individualized care. [1,2]

Treatment and therapy

Infants may need feeding support and therapy. Later nutritional planning and predictable, supervised food access protect health without shaming the child. Endocrine care may include growth hormone when appropriate, with required monitoring. Sleep assessment, developmental therapies, communication support, and educational accommodations are important. Medicines and hormone treatments require specialist selection and follow-up. [1,2,3]

Support at home

Follow the current nutrition plan rather than applying blanket food restriction. Establish predictable meals and consistent food-access arrangements across caregivers while preserving dignity. Prepare for changes with simple explanations and visual routines. Support enjoyable movement and monitor sleep, constipation, and other discomforts with the care team.

When to seek help

Obtain urgent assessment for severe abdominal pain or distension, unusual vomiting, breathing problems, marked lethargy, or inability to feed. Infants with inadequate intake need prompt clinical review.

Questions for your care team

- How should nutrition change at this developmental stage?
- Which endocrine and sleep monitoring is needed?
- How can caregivers use a consistent, respectful food-access plan?



Podcast preparation

Use this outline to develop a later podcast script.

Suggested opening

Welcome to Mariel Early Intervention. Today we discuss prader-willi syndrome. Prader-Willi syndrome is a genetic condition with changing needs across childhood. This is general education; each child needs individualized care.

Opening: Define the topic and its developmental context.

Understanding: Explain causes and signs without inviting self-diagnosis.

Assessment: Describe evaluation and distinguish screening from diagnosis.

Support: Discuss treatment, therapy, and practical family supports.

Closing: Explain urgent signs and one clear next step.

Misconception to address: Myth: food seeking is simply misbehavior. Altered appetite regulation requires structured, compassionate support.

Before recording: update clinical references and local service details, arrange clinical review, and use fully consented or fictional examples.

Terms in plain language

- Hyperphagia: an unusually strong or persistent drive to eat.
- Hypotonia: reduced resting muscle tone.
- Endocrine care: assessment and treatment of hormone-related health needs.

Sources and further reading

Sources accessed 3 October 2026. Numbered references support the clinical overview. Treatment availability and referral routes vary by country.

[1] Prader-Willi syndrome: MedlinePlus Genetics

<https://medlineplus.gov/genetics/condition/prader-willi-syndrome/>

[2] What are the treatments for Prader-Willi syndrome (PWS)?

<https://www.nichd.nih.gov/health/topics/prader-willi/conditioninfo/treatments>

[3] AAP Releases New Guidance on Faltering Weight in Children

<https://www.healthychildren.org/English/news/Pages/aap-issues-new-guidance-on-faltering-weight-in-children.aspx>