Understanding Prader-Willi Syndrome

Prepared by

FOUNDATION FOR PRADER-WILLI RESEARCH

What is Prader-Willi syndrome?

FOUNDATION FOR PRADER-WILLI RESEARCH

What is Prader-Willi syndrome?

- Prader-Willi syndrome (PWS) is a rare genetic disorder affecting approximately 1 in 15,000 people.

- PWS is a life-threatening medical disorder caused by loss of active genetic material on the paternal copy of chromosome 15.

Who is at risk?

Who is at risk?

- PWS occurs randomly. There is nothing that a pregnant woman does, or does not do, that causes PWS.

- PWS affects males and females and people of all races and ethnicities equally.

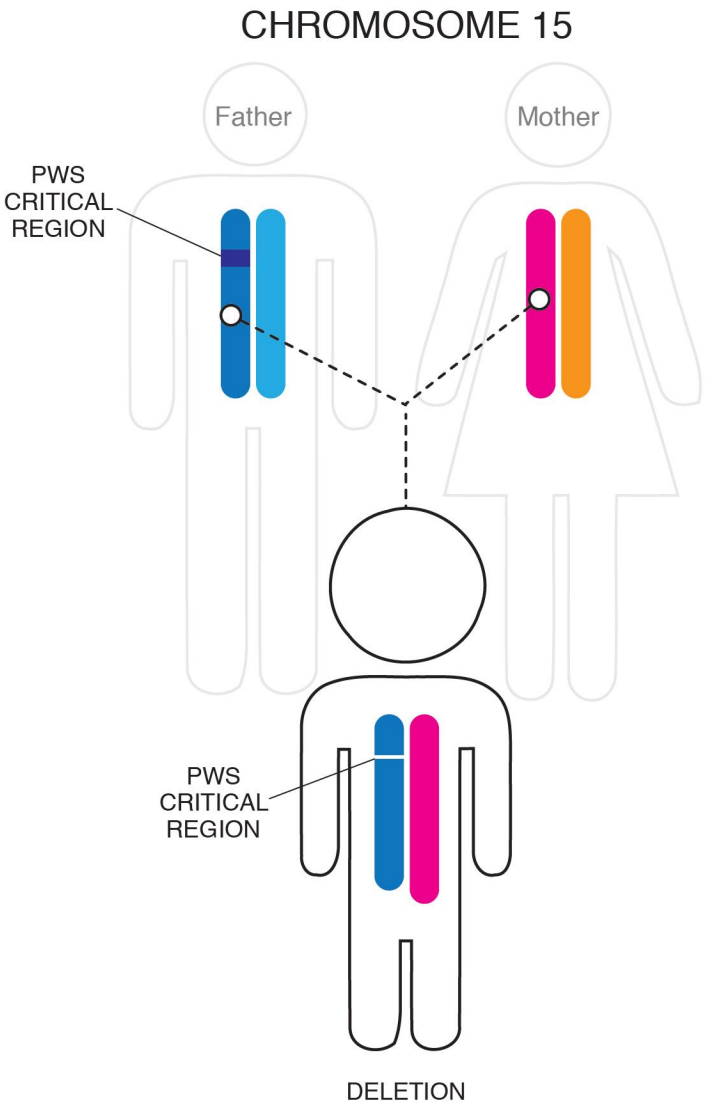
What causes Prader-Willi syndrome?

What causes Prader-Willi syndrome?

PWS by Deletion - Most often, part of the chromosome 15 that was inherited from the person's father is missing, or deleted, in this critical region. This small deletion occurs in approximately 70% of cases.

PWS by UPD - Another 30% or so of cases occur when an individual inherits two chromosome 15s from their mother and none from their father. This scenario is termed maternal uniparental disomy (UPD).

PWS by Methylation Defect (Imprinting Mutation) - In a very small percentage of cases (1-3%), a small genetic mutation in the Prader-Willi region causes the paternal chromosome 15 genetic material (although present) to be inactive.

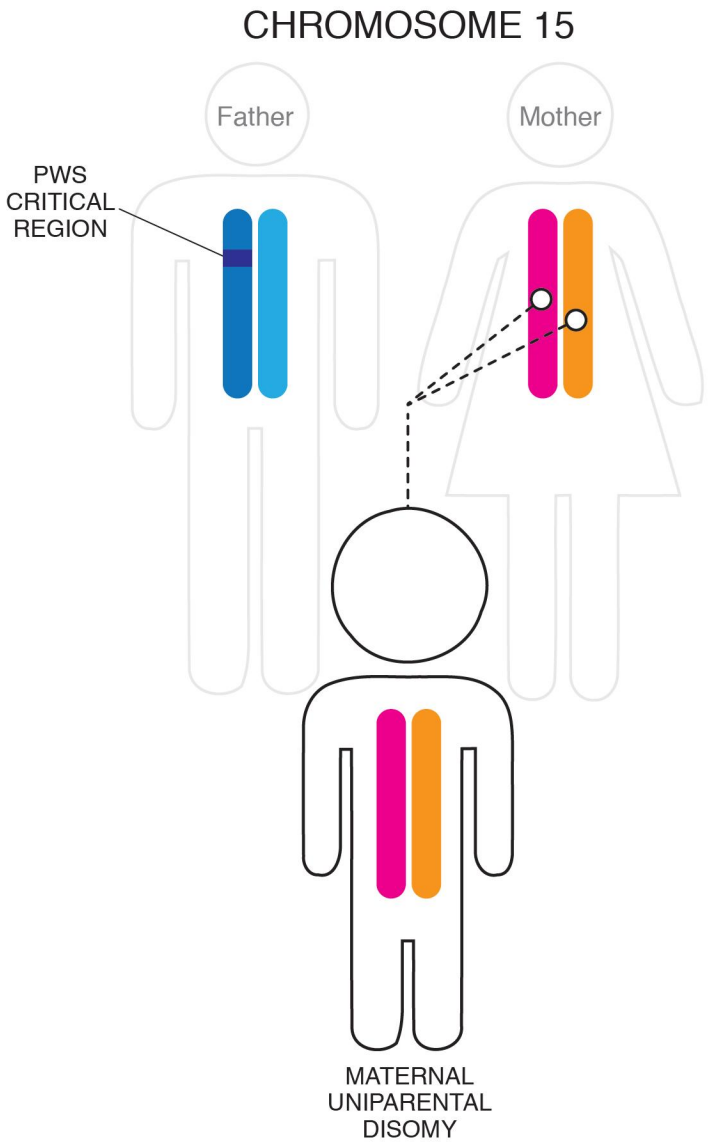


What causes Prader-Willi syndrome?

PWS by Deletion - Most often, part of the chromosome 15 that was inherited from the person's father is missing, or deleted, in this critical region. This small deletion occurs in approximately 70% of cases.

PWS by UPD - Another 30% or so of cases occur when an individual inherits two chromosome 15s from their mother and none from their father. This scenario is termed maternal uniparental disomy (UPD).

PWS by Methylation Defect (Imprinting Mutation) - In a very small percentage of cases (1-3%), a small genetic mutation in the Prader-Willi region causes the paternal chromosome 15 genetic material (although present) to be inactive.

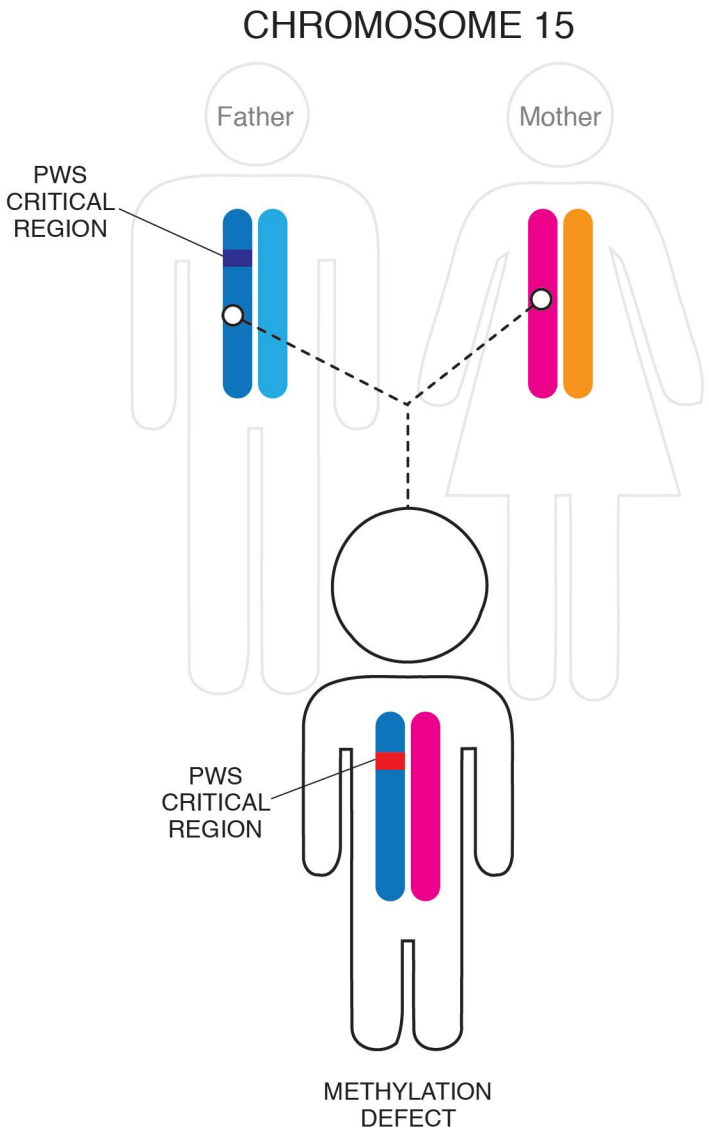


What causes Prader-Willi syndrome?

PWS by Deletion - Most often, part of the chromosome 15 that was inherited from the person's father is missing, or deleted, in this critical region. This small deletion occurs in approximately 70% of cases.

PWS by UPD - Another 30% or so of cases occur when an individual inherits two chromosome 15s from their mother and none from their father. This scenario is termed maternal uniparental disomy (UPD).

PWS by Methylation Defect (Imprinting Mutation) - In a very small percentage of cases (1-3%), a small genetic mutation in the Prader-Willi region causes the paternal chromosome 15 genetic material (although present) to be inactive.



What challenges do people with PWS face?

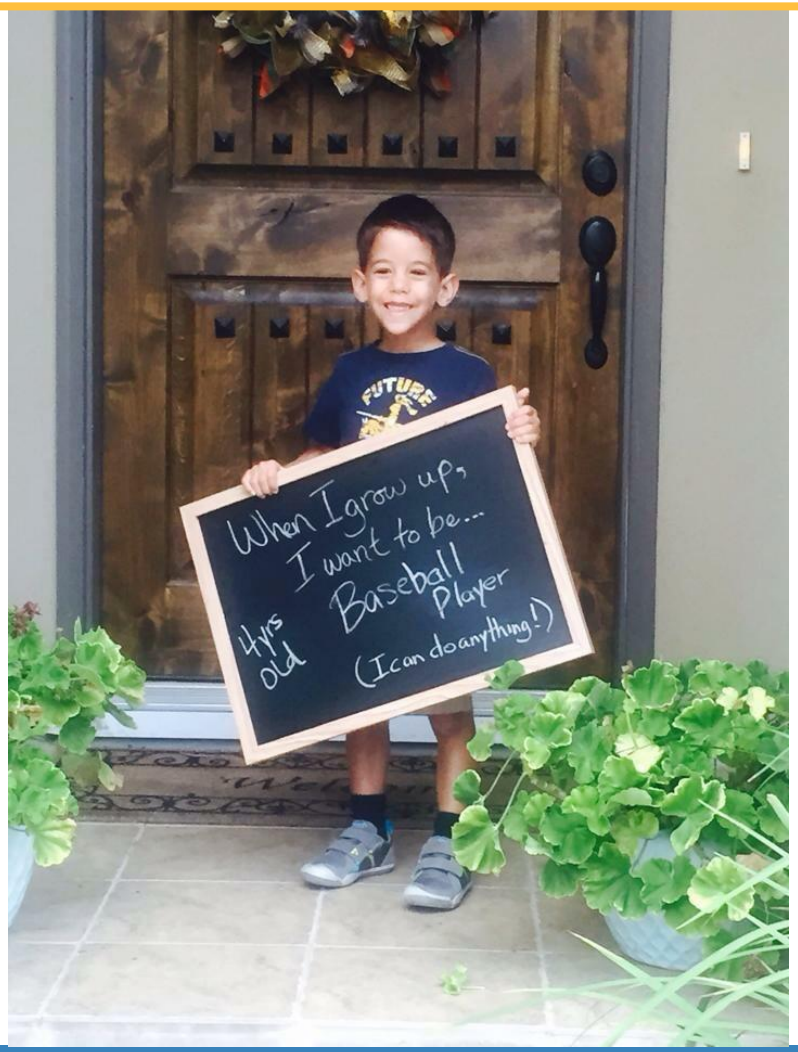
What challenges do people with PWS face?

The hallmark symptom of PWS is hyperphagia, or continuous, extreme hunger. A person with PWS never feels full. Exacerbating this problem, those with PWS have a slow metabolism and need only a fraction of the calories of their typical peers. The result is easy weight gain. PWS is recognized as the leading genetic cause of life-threatening obesity in children.

What challenges do people with PWS face?

Nearly every system in the body is impacted by a PWS diagnosis. PWS affects:

- Hormones
- Body composition
- Muscle strength
- Appetite
- Behavior
- Cognition and learning
- Temperature regulation
- Pain tolerance
- Sleep patterns



Is there a cure for Prader-Willi syndrome?

Is there a cure for Prader-Willi syndrome?

Currently, there is **no cure** for Prader-Willi syndrome.

For many individuals affected by the disorder, the elimination of some of the most difficult aspects of the syndrome, such as the insatiable appetite and obesity, would represent a significant improvement in quality of life and the ability to live independently.

Advancing Research

The Foundation for Prader-Willi Research is advancing research toward understanding and treating specific aspects of the syndrome, with the goal of an eventual cure for PWS.

The Foundation for Prader-Willi Research

Founded by mothers of children with PWS, the Foundation for Prader-Willi Research recognizes the urgency for finding treatments for the many challenges our loved ones face. We prioritize innovative research that will help develop therapies and positively impact the lives of all individuals with PWS.

FOUNDATION FOR PRADER-WILLI RESEARCH
Working Toward an Independent Future

What Can You Do?

Learn more about FPWR's research programs and get involved at **www.fpwr.org**

What Can You Do?

Donate to support PWS research at www.fpwr.org/donate-friends

Thank you!

Together we can create a future of hope!