PRADER-WILLI SYNDROME ASSOCIATION
Still hungry for a cure.

PRADER-WILLI SYNDROME FACT SHEET

WHO has Prader-Willi syndrome (PWS)?

Anyone can be born with Prader-Willi syndrome (PWS). 'Acquired PWS' can result later in life from brain trauma.

WHAT is Prader-Willi syndrome?

PWS is a complex genetic disorder affecting appetite, growth, metabolism, cognitive function and behavior. It is typically characterized by low muscle tone, short stature, incomplete sexual development, cognitive disabilities, problem behaviors, and the hallmark characteristics—involuntary and uncontrollable chronic feelings of hunger and a slowed metabolism that can lead to excessive eating and life-threatening obesity. Those who have PWS need intervention and strict external controls, including padlocking access to food, to maintain normal weight and to help save their lives.

WHEN does Prader-Willi syndrome occur?

It is estimated that PWS occurred in one in 12,000 to 15,000 births. Although considered a "rare" disorder, PWS is one of the most common conditions seen in genetics clinics and is the most common genetic cause of obesity that has been identified.

WHERE is Prader-Willi syndrome found?

PWS is found in people of both sexes and all races worldwide.

WHY does Prader-Willi syndrome occur?

Most cases of PWS are attributed to a spontaneous genetic error that occurs at or near the time of conception for unknown reasons. In a very small percentage of cases (2 percent or less), a genetic mutation that does not affect the parent is passed on to the child, and in these families more than one child may be affected. A PWS-like disorder, 'Acquired PWS', can also develop after birth if the hypothalamus portion of the brain is damaged through injury or surgery.

HOW does Prader-Willi syndrome work?

The occurrence of PWS is due to lack of several genes on one of an individual's two chromosome 15s—the one normally contributed by the father. In most cases, there is a deletion—the critical genes are somehow lost from the chromosome. In most of the remaining cases, the entire chromosome from the father is missing and there are instead two chromosome 15s from the mother (uniparental disomy). The critical paternal genes lacking in people with PWS have a role in the regulation of appetite. This is an area of active research in a number of laboratories around the world, since understanding this defect may be very helpful not only to those with PWS but to understanding obesity in otherwise normal people.

People with PWS have a flaw in the hypothalamus part of their brain, which normally registers feelings of hunger and satiety. While the problem is not yet fully understood, it is apparent that people with this flaw never feel full; they have a continuous urge to eat that they cannot learn to control. To compound this problem, people with PWS need less food than their peers without the syndrome because their bodies have less muscle and tend to burn fewer calories. The hypothalamus is also related to rage and other behavioral problems.

WHAT does Human Growth Hormone do for people with Prader-Willi syndrome?

Use of Human Growth Hormone is becoming standard care for those with PWS, when prescribed with appropriate precautions by an experienced endocrinologist. Human Growth Hormone can help with height, weight, body mass, strength, and agility, among other areas. It may also help with cognitive development.

Reports have indicated dramatic increase in growth rate (especially in the first year of treatment) and a variety of other effects, including improved body composition (higher muscle mass, lower fat mass); improved weight management; increased energy and physical activity; improved strength, agility and endurance; and improved respiratory function

WHO to contact for more information:

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