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# An introduction to Prader-Willi Syndrome

## What is Prader-Willi Syndrome?

Prader-Willi Syndrome (PWS) is a lifelong condition and although lots of research is being carried out, to date there is no cure. There are no known reasons for the genetic accident that causes this condition, but we are learning more about PWS everyday, and there is hope. Hope that, through increased knowledge, we can help all people with this condition to live a fuller life despite their limitations.

Prader-Willi Syndrome (PWS) is a rare and very complex non-inherited genetic disorder in which several genes on the 15th chromosome are deleted or unexpressed. PWS was first described by Swiss doctors Andrea Prader, Alexis Labhart and Heinrich Willi in 1956 based on the clinical characteristics of nine children they examined.

In recent years, the syndrome has been genetically characterized as an abnormality of chromosome 15, and definitive diagnosis is now based on genetic testing.

Chromosome 15 is one of the 23 pairs of chromosomes in humans. People normally have 2 copies of this chromosome.

Chromosome 15 contains between 700 and 900 genes. Angelman Syndrome, PWS and now Autism are all related to Chromosome 15.

Every case of PWS is due to the baby failing to receive active genes from a specific section of chromosome 15, there are 3 different ways that this can happen:

**1) Paternal deletion** – about 70% of all cases of PWS. In the most common form of PWS, part of the chromosome 15 inherited from the father – the part containing the PWS critical genes –is missing. Usually a deletion happens for no known reason.

**2) Maternal uniparental disomy (UPD)** – about 25% of cases. In this less common form of PWS, the baby inherits both copies of chromosome 15 from the mother.

**3) Imprinting defect** – less than 5% of cases. Further testing and genetic counselling are especially important for families who have a child with an imprinting defect.

Symptoms associated with Prader-Willi Syndrome (PWS) are believed to be caused, in part by a defect in the hypothalamus; an important supervisory centre in the brain that controls metabolism of fats and carbohydrates, the development of muscle tone, the regulation of the sleep/wake cycle, body temperature, blood pressure, heartbeat, the expression of emotions, and many more functions of the body.[1]

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# An introduction to Prader-Willi Syndrome (cont'd)

## Characteristics of Prader-Willi Syndrome

Prader Willi Syndrome (PWS) was first described in 1956 by Swiss doctors, Prof. A. Prader, Dr. A. Labhart and Dr. H. Willi, who recognised the condition as having unique and clearly definable features. These features are:-

**Hypotonia:**

Weak muscle tone, and floppiness at birth.

**Hypogonadism:**

Immature development of sexual organs and other sexual characteristics.

**Obesity:**

Caused by excessive appetite and overeating (hyperphagia), and a decreased calorific requirement owing to low energy expenditure levels. (Obesity is not normally a feature of those whose food intake is strictly controlled.)

**Central nervous system and endocrine gland dysfunction:**

Causing varying degrees of learning disability, short stature, hyperphagia, somnolence, and poor emotional and social development.

**Physical features:**

Many people with PWS also exhibit characteristic facial and other physical features. These include: almond-shaped eyes, a narrow forehead (measured across), a down-turned mouth with a triangular-shaped upper lip, and small hands and feet.

**Poor large muscle strength:**

Often coupled with poor coordination and balance. Muscle tone can be improved with appropriate therapy and exercise. Small muscle strength is usually better.

**Learning difficulties:**

Most people with PWS have borderline or moderate learning difficulties. Some people have recorded IQs of 90 or above, whilst a minority have severe learning difficulties. The average IQ is around 60 - 70, although individuals may sometimes find it difficult to perform at their IQ level, as emotional and social skills are often less developed. Reading and writing skills are usually considerably better than number skills and abstract thinking.

**Short stature and abnormal body composition:**

Short stature and small hands and feet are a feature of most people with PWS, and lack of growth hormone (GH) and delayed or absent puberty are probably contributory to this. Growth hormone therapy improves final height, physical strength, agility and respiratory muscle function, as well as increasing the overall muscle mass and reducing the fat mass in the body. Growth hormone therapy will not necessarily help with obesity, nor is there any evidence to show that it decreases the feelings of hunger.[2]


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