

## PWS in adulthood

If untreated, adults are usually shorter than expected for the family, significantly overweight, with a small head circumference and small hands and feet. Excessive eating and obesity pose a huge risk to their health. In addition to the increased risk of diabetes, they have a higher propensity to respiratory and skin infections, particularly at a younger age, significantly increased risk of cardiovascular disease, obstructive sleep apnoea and acute, life threatening complications due to gastric (stomach) distension.

## Treatment of PWS

Unfortunately, there is no cure for PWS, but once the diagnosis is made they will require input from a number of specialists. Management of the feeding is very important in the first year of life to maintain adequate weight gain, but with the onset of hyperphagia, an appropriate dietary plan as well as physical activity is necessary to prevent obesity.

Treatment with growth hormone has shown good effects on achieving normal height, increasing mobility and helps weight management. Growth hormone treatment may increase the cognitive and language skills in young children and mental speed and

motor performance in adults. These interventions significantly change the disease course and improve the outcome and quality of life.

## Further Information and support is available from:

Prader-Willi Syndrome Association (UK)  
125a London Road,  
Derby,  
DE1 2QQ  
Tel: 01332 365 676  
Fax: 01332 360401  
Email: [admin@pwsa.co.uk](mailto:admin@pwsa.co.uk)  
Website: <http://pwsa.co.uk>

## Acknowledgment

With thanks to Guy's and St Thomas' NHS Foundation Trust, London.

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**Birmingham Women's NHS Foundation Trust**  
Edgbaston, Birmingham, B15 2TG  
Telephone: 0121 472 1377  
Fax: 0121 627 2602

Reference Number: GG 23  
Author: Dr Jenny Morton  
Consultant Clinical Geneticist

Reviewed: June 2011 Next Review: June 2014

Birmingham Women's   
NHS Foundation Trust

# Prader-Willi Syndrome

An information leaflet for  
parents and families

If you need more advice about any aspect  
of Prader-Willi syndrome please contact:

**Clinical Genetics Unit**  
Birmingham Women's  
NHS Foundation Trust  
Mindelsohn Way  
Edgbaston  
Birmingham  
B15 2TG

Telephone: 0121 627 2630

Fax: 0121 627 2618

Email: [Clinicalgenetics.info@bwhct.nhs.uk](mailto:Clinicalgenetics.info@bwhct.nhs.uk)

## What is Prader-Willi syndrome

Prader-Willi Syndrome (PWS) is a rare genetic disorder. Affected children are noted to have short stature, feeding difficulties in infancy followed by obsessive eating leading to severe obesity, motor delay, mild to moderate learning difficulties and behavioural problems.

## What are chromosomes and genes?

Chromosomes are condensed strings of DNA. We have 46 chromosomes in most cells in our body, arranged in pairs. We normally inherit one of each of the pairs from our mother and one from our father.

Each chromosome carries hundreds of genes. Genes are unique DNA sequences that determine a particular characteristic or function. We have more than 25,000 different genes. The combination of the genes we inherit makes us all individual.

## What causes PWS?

We need a copy of chromosome 15 from each parent. If a specific part of chromosome 15, inherited from the father, is not present or not working correctly (known as loss of paternal expression) then PWS occurs.

Prader-Willi Syndrome can be caused by one of several different mechanisms:

- *Deletion of part of the paternal chromosome 15.* This is the most common cause of Prader-Willi syndrome found in approximately 70% cases.
- *Chromosome 15 maternal Uniparental Disomy (UPD).* This means that instead of there being a copy of chromosome 15 from each parent, both copies are inherited from the mother. This is found in about 25% of patients.
- *Alteration of the imprinting centre* which makes sure that the region on the paternal chromosome 15 involved in Prader-Willi Syndrome is properly represented (expressed). About 4% of cases are caused by this mechanism.
- *Chromosome rearrangement* involving the paternal chromosome 15.

Virtually all cases of Prader-Willi Syndrome can be confirmed by genetic tests.

## Will it happen again?

Most cases of Prader-Willi Syndrome are a "one off" and the chance of having a further child with this condition would be small. A very small proportion of cases may carry an increased risk of recurrence and this can be discussed at the genetics clinic. Prenatal diagnosis (testing a baby during pregnancy) may be available. This can be discussed at the genetics clinic.

## PWS in infancy and childhood

There may be a history of poor fetal movement during pregnancy, but a newborn baby with Prader-Willi is usually of average size and looks healthy. However they have low muscle tone, poor ability to suck and significant feeding difficulties meaning they may have difficulty putting weight on and growing normally.

Hyperphagia (overeating) and rapid weight gain develops between the age of one and six years. Children constantly feel hungry, and can go to extreme lengths to obtain food. This leads to significant obesity making physical activity difficult.

Most children with PWS take double the normal times to achieve their developmental milestones and on average start walking at around the age of 2 years. Children with PWS can have good expressive and receptive language but may also have complex learning difficulties and may require special education. Sexual development and puberty may be delayed or impaired and may require treatment.

Behavioural problems such as attention deficit hyperactivity disorder, temper tantrums, obsessive compulsive disorder or autistic spectrum disorder are common. These behavioural patterns tend to worsen with age and increased body mass index, but become less troublesome in older adults.