PWS in adulthood
If untreated, adults are usually shorter than expected for the family and are significantly overweight, with small head circumference and small hands and feet. Excessive eating and obesity pose a significant risk to their health. In addition to the increased risk of diabetes, individuals have a higher risk of respiratory and skin infections, particularly at a younger age; significantly increased risk of cardiovascular disease; obstructive sleep apnoea (breathing problems during sleep) and acute, life threatening complications due to gastric (stomach) distension. A person with PWS may have characteristic facial features that can be recognised by a Geneticist.

Treatment of PWS
Unfortunately, there is no cure for PWS, but once the diagnosis is made input can be given from a number of specialists. Management of feeding is very important in the first year of life to maintain adequate weight gain, but with the onset of hyperphagia (overeating), an appropriate dietary plan as well as physical activity are necessary to prevent or minimise obesity.

Treatment with growth hormone has shown good effects on achieving normal height and increasing mobility, and helps with weight management. The treatment may also 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.

Feedback
We appreciate and encourage feedback. If you need advice or are concerned about any aspect of your care or treatment please speak to a member of staff or contact the Patient Advice and Liaison Service (PALS):

Freephone: 0800 183 0204
From a mobile or abroad: 0115 924 9924 ext. 65412 or 62301
E-mail: pals@nuh.nhs.uk
Letter: NUH NHS Trust, c/o PALS, Freepost NEA 14614, Nottingham NG7 1BR

www.nuh.nhs.uk

Further information
Prader-Willi Syndrome Association UK
Suite 4.4, Litchurch Plaza, Litchurch Lane, Derby DE24 8AA

Tel: 01332 365676
Email: admin@pwsa.co.uk
www.pwsa.co.uk

Adapted from leaflet by Clinical Genetics at Guy’s and St Thomas’ NHS Foundation Trust.
What is Prader-Willi syndrome?
Prader-Willi syndrome (PWS) is a rare genetic disorder. Affected children tend to have:
- Short stature (below average height)
- Feeding difficulties in infancy followed by obsessive eating at a few years old
- Severe obesity
- Low muscle tone (hypotonia)
- Motor delay (slow development in movement control skills)
- Mild to moderate learning difficulties and behavioural problems.

What are chromosomes and genes?
Chromosomes are thread-like structures that are made up of tightly packaged DNA. We have 46 chromosomes in most cells in our body, arranged in pairs. We normally inherit one of each of the pairs of chromosomes from our mother and one from our father. Each chromosome carries hundreds of genes. Genes are a unique DNA sequence that acts as instructions that tell our cells how to function and what characteristics to express. We have about 20,000 different genes. The combination of the genes we inherit makes each of us an individual.

What causes PWS?
We inherit one copy of chromosome 15 from both parents. 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 PWS and is found in approximately 70% of cases.
- Chromosome 15 maternal uniparental disomy (UPD): This means that instead of inheriting 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 correctly represented (expressed). About 4% of cases are caused by this mechanism.
- Complex rearrangement involving the paternal chromosome 15.

Virtually all cases of PWS can be confirmed by genetic tests.

PWS in infancy and childhood
There may be a history of poor fetal movement during pregnancy, but a newborn baby with PWS 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 on weight 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 time to achieve their developmental milestones and on average start walking at around two years of age.

Children with PWS can have good expressive and receptive language but also have complex learning difficulties and may require special education. Sexual development may not occur as normal and puberty may be delayed or impaired.

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