

# Prader Willi Syndrome

## Definition of Prader Willi Syndrome

Prader-Willi syndrome is a distinct condition characterised by neurological impairments causing an altered pattern of growth and development with a unique hyperphagia (over-eating). This preoccupation with food and accompanying compulsion to eat can cause extreme obesity with premature death. Early intervention with a dietitian and behavioural psychologist to prevent excessive weight gain in childhood is crucial.

## Cause of Prader Willi Syndrome

Prader-Willi syndrome is caused by a lack of a functioning paternally-inherited region of chromosome 15q11-13. Prenatal diagnosis is possible, although risk of recurrence is very low (except when caused by an imprinting anomaly). The condition is mostly sporadic, affecting both males and females. Estimates of the incidence vary between 1/8,500 to 1/38,000 births.

## Presentation

### Presentation in Neonates and Infant

- Neonates may have severe hypotonia (floppiness) with a weak suck, often necessitating nasogastric feeding.
- Severe delay in motor development is common.
- Hypogonadism in boys is common.

### Presentation in Children

- The physical phenotype, characterised by deep-set almond-shaped eyes, narrow forehead and small hands and feet, is usually recognisable during the second year of life.
- There is a delay in sitting and crawling, but the severe hypotonia improves and most children start walking independently between two and three years.
- Young children may present with mild to moderate global developmental delay.
- Learning difficulties associated with a mild intellectual disability are usually obvious by school entry.
- Preoccupation with food usually becomes evident during the pre-school years.
- Food-seeking behaviours are often evident by school age.
- The onset of obesity can be subtle and occur without any obvious change in eating behaviour or increased calorific intake.
- Short stature usually develops as childhood and adolescent growth spurts may be delayed or absent.
- The behavioural phenotype may become evident during childhood with exacerbations during adolescence.
- Delayed or incomplete puberty is common.

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## Presentation in Adults

The diagnosis should be considered in patients with short stature who demonstrate a behavioural phenotype which includes:

- Obesity with food-seeking behaviour. Obesity increases the risk of diabetes, coronary artery disease, cor pulmonale, sleep apnoea and premature death.
- Low energy expenditure.
- Severe delays in adaptive behaviour.
- Limited speech.
- Perseverative behaviours.
- Reduced Physical Activity. Hypotonia with reduced muscle bulk and strength persists into adult life and contributes to poor posture and reduced stamina for physical activities.

## **Diagnosis**

Prader-Willi syndrome can be diagnosed with a blood test using methylation analysis. This test detects the deletion, uniparental disomy (which are sporadic with no risk of recurrence) and imprinting mechanisms (which can have a recurrence rate of up to 50%). If the test is negative and there is a high index of suspicion referral to a geneticist is indicated to investigate for rare anomalies.

Other diagnostic tests used are FISH (fluorescence in situ hybridisation) to detect deletions and 15 PCR to detect paternal disomy.

## **Support Association**

Prader-Willi Syndrome Association of Australia  
Telephone: (02) 4946 9001  
<http://www.pws.org.au>

The material in this sheet has been adapted from the Therapeutic Guidelines book 'Management Guidelines for People with Developmental and Intellectual Disabilities' and updated from the 2005 version, Management Guidelines – Developmental Disability' which can be consulted for more detailed information.

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