

## Prader-Willi syndrome

Prader-Willi syndrome is a rare genetic disorder that affects development and growth. Estimates of its incidence vary; around one in 10,000 to 20,000 children are born with the syndrome, with females slightly more prone than males.

There is no cure for the condition, but professional health care from a range of specialists can improve the child's quality of life.

### Symptoms and signs

Typically, the affected child is unusually floppy at birth and has feeding difficulties and a weak cry. Males frequently have undescended testes and may have underdeveloped genitalia. Around half of all children with Prader-Willi syndrome will have fair skin, blonde hair and blue eyes, regardless of what their family members look like.

The affected child is delayed in all aspects of development, reaching developmental milestones – such as sitting, crawling and walking – later than other children. The average IQ is around 70, but the degree of intellectual disability will differ for each child.

By five years of age, a feature of Prader-Willi syndrome is the child's excessive appetite, which often leads to obesity if not managed carefully with dietary and behavioural control.

### The cause is genetic

Humans have 23 pairs of chromosomes, with one set inherited from each parent. Research indicates that missing genes on chromosome 15, contributed by the father, cause Prader-Willi syndrome. The loss can happen in four ways, including:

- The father's chromosome 15 is altered – usually, genes are deleted. This is the most common cause, accounting for between 60 and 70 per cent of cases.
- The baby inherits two of the chromosomes from the mother, and none from the father. This happens in about 25 to 30 per cent of cases.
- A translocation occurs, which means some of the genes on chromosome 15 get shuffled around or swapped with genes from other chromosomes.
- The father's chromosome 15 is intact, but the genes in the Prader-Willi region do not work properly.

### Affected individuals are prone to obesity

At birth, the child has poor sucking ability and may not grow at the expected rate. However, this changes remarkably by the time the child is one to four years of age. Prader-Willi syndrome is associated with an excessive appetite, which means that the child is prone to obesity. This is a challenge for a child with Prader-Willi syndrome and their family, but it can be managed by dietary and behavioural measures.

### Other associated problems

A child with Prader-Willi syndrome is prone to a range of associated health and behavioural problems as they get older. Some of these problems may include:

- Obsessive and compulsive behaviours, such as picking at the skin
- Eye problems, such as nearsightedness
- Short stature, often due to growth hormone deficiency

- Delayed onset of puberty
- Scoliosis (sideways curves in the spine)
- Kyphosis (exaggerated hump in the spine)
- Delayed or absent menstrual periods in girls
- Abnormally small penis in boys
- Diabetes, triggered by obesity
- Osteoporosis (weakened bones that are prone to fracturing)
- Teeth problems, including soft enamel and tooth grinding
- Sleep apnoea (periods of breathing cessation during sleep)
- Problems with short term memory
- Temper tantrums.

## Diagnosis methods

Prader-Willi syndrome is diagnosed by physical examination and blood tests to check for problems with chromosome 15.

## Treatment options

There is no cure for Prader-Willi syndrome and no means of prevention. Treatment aims to ease some of the associated problems. Depending on the needs of the individual, some of the treatment options may include:

- Strict supervision of diet. To date, there are no medical means of curbing appetite
- Plenty of physical activity to help maintain the child's body weight within the normal range
- Growth hormone treatment to overcome the hormone deficiency that contributes to the child's short stature
- Hormone therapy to increase muscle mass
- Hormone therapy to boost inadequate sex hormone levels
- Medications to help control any obsessive and compulsive behaviours
- Orthopaedic treatment for scoliosis or kyphosis
- Appropriate prescription eye glasses
- Specialist care from a range of health care professionals.

## Specialist care is important

A child with Prader-Willi syndrome will benefit enormously from specialist care. Health professionals often include:

- General practitioner
- Paediatrician
- Dietitian
- Physiotherapist
- Speech therapist
- Dentist
- Optician
- Behavioural psychologist.

## Where to get help

- Your doctor
- Prader-Willi Syndrome Association of Australia.

## Things to remember

- Prader-Willi syndrome is a rare genetic disorder affecting development and growth.
- A child with Prader-Willi syndrome has an excessive appetite, which often leads to obesity.
- Other characteristics include short stature and intellectual disability.
- Treatment from health care professionals leads to improved quality of life.

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Genetic Health Services Victoria

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