

Prader-Willi syndrome

Prader-Willi syndrome is a rare genetic disorder, which 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. Typically, the affected child will be small and unusually floppy at birth, with abnormal limb development and disproportionately small hands and feet. 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 will still reach developmental milestones - such as sitting, crawling and walking - but at a much later stage than normal. The average IQ is around 70, but the degree of intellectual disability will differ from one individual to the next. The poor muscle tone that makes sucking difficult for the baby will later contribute to speech development problems. A feature of Prader-Willi syndrome is the child's voracious and insatiable appetite, which often leads to obesity. 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

Sometimes, signs of Prader-Willi syndrome may be evident during the pregnancy. Some of these signs may include weak foetal movements and small size for gestational age. The symptoms of Prader-Willi syndrome in a newborn baby may include:

- Floppiness, with weak muscle tone
- Small size compared to normal development
- Narrow skull
- Defects of the long bones
- Disproportionately small hands and feet
- Weak cry
- Poor sucking ability
- Smaller than normal genitals, which can include undescended testicles in male infants.

The cause is genetic

Humans have 23 pairs of chromosomes, 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 unusable.

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 insatiable appetite, so that the child will eat virtually everything, and may be so obsessed by food they will go to any lengths to get it. Coupled with the unusually slow metabolism characteristic of the syndrome, this means that the child is prone to obesity. The hypothalamus is the brain structure responsible for regulation of appetite. It is thought that the hypothalamus in a child with Prader-Willi syndrome is dysfunctional. This is a lifelong problem.

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:

- Eye problems, such as nearsightedness

- Stunted growth
- 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
- Obsessive and compulsive behaviours, such as picking at the skin.

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 bodyweight 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-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. The professionals often associated include:

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

Where to get help

- Your doctor
- Prader-Willi Syndrome Association of Victoria Tel. (03) 9735 5199
- Prader-Willi Syndrome Association of Western Australia Tel. (08) 9375 8104
- Prader-Willi Syndrome Clinic, Royal Children's Hospital ([hyerlink as below](#))- or you could use the RCH phone number.

Things to remember

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

This page has been produced in consultation with, and approved by:

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