Prader-Willi syndrome

Summary

- Prader-Willi syndrome is a rare genetic disorder affecting development and growth.
- A child with Prader-Willi syndrome has an excessive appetite, which can lead to obesity if not properly managed.
- Other characteristics include short stature and intellectual disability.
- Treatment from healthcare professionals leads to improved quality of life.

Prader-Willi syndrome is a rare genetic disorder that affects development and growth. It is thought that around one in 10,000 to 20,000 children are born with the syndrome, with females slightly more likely to have the condition than males.

There is no cure for Prader-Willi syndrome, but professional health care from a range of specialists can improve the child’s quality of life.

Symptoms of Prader-Willi syndrome

Typically, a child with Prader-Willi syndrome is unusually floppy at birth and has feeding difficulties and a weak cry. Males frequently have testes that haven’t moved to the scrotum (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. Children with Prader-Willi syndrome are delayed in all aspects of development, reaching developmental milestones – such as sitting, crawling and walking – later than other children. The average IQ of a child with Prader-Willi syndrome 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 excessive appetite, which can lead to obesity if not carefully managed.

Causes of Prader-Willi syndrome

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. This can happen in four ways:

- 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.

Obesity and Prader-Willi syndrome

At birth, children with Prader-Willi syndrome have poor sucking ability and may not grow at the expected rate. However, this changes remarkably by the time they are one to four years of age.

From this age, Prader-Willi syndrome is associated with an excessive appetite, which means that children with Prader-Willi syndrome are prone to obesity. This is a challenge for children with Prader-Willi syndrome and their families, but it can be managed by dietary and behavioural measures.

Problems associated with Prader-Willi syndrome

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

- obsessive and compulsive behaviours, such as picking at the skin
- eye problems, such as short-sightedness
- 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
- type 2 diabetes
- osteoporosis (weakened bones that are prone to fracturing)
- teeth problems, including soft enamel and tooth grinding
- sleep apnoea (breathing stops for a period of time during sleep)
- problems with short-term memory
- temper tantrums.

Diagnosis of Prader-Willi syndrome

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

Treatment for Prader-Willi syndrome

There is no cure for Prader-Willi syndrome and it cannot be prevented. Treatment aims to ease some of the associated problems. Depending on the needs of the person, some of the treatment options may include:

- strict supervision of diet (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
- medication to help control any obsessive and compulsive behaviours
- orthopaedic treatment for scoliosis or kyphosis
- appropriate prescription eye glasses
- specialist care from a range of healthcare professionals.

Specialist care is important

Children with Prader-Willi syndrome will benefit enormously from specialist care. Health professionals involved in their care often include:

- general practitioner (doctor, or GP)
- paediatrician (a doctor who specialises in treating children)
- dietitian
- physiotherapist
- speech therapist
- dentist
- optician
- behavioural psychologist.

Where to get help

- Your doctor (GP)
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