



Prader–Willi syndrome

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What is it?

Prader-Willi syndrome is a genetic disorder. Individuals suffering from the syndrome have specific characteristics such as hypotonia (low muscle tone), feeding difficulties and incomplete sexual development in the neonatal period, and obesity, short stature, mental impairment and behavioural problems later on. The condition was first described in the medical literature in 1956 by three doctors: A. Prader, H. Willi and A. Labhart, hence the name. The prevalence is between 1 in 10,000 and 1 in 15,000 live births. This syndrome occurs equally in males and females and is found in individuals of all races.

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What are the characteristics of Prader-Willi syndrome?

Numerous characteristics have been described in the literature, but it is important to remember that the clinical picture can vary and changes with age.

Main characteristics:

1. In the neonatal period:

- Hypotonia (floppiness or low muscle tone) with weak or absent cry
- Feeding difficulties requiring special assistance in most cases (gastric tube feeding)
- Incomplete sexual development, e.g. undescended testicles
- Characteristic facial features, e.g. almond shape eyes, narrow forehead, etc.
- History of decreased fetal movements with high number of breech presentation

2. In the childhood period:

- Increased appetite and excessive weight gain from 2 to 3 years of age
- Delay in attaining early developmental milestones
- Behavioural problems with temper tantrums mostly centered round a constant craving for food
- Intellectual impairment with learning problems
- Short stature
- Lack of normal sexual development
- Skin picking

Numerous other features have also been described.

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What is the cause of Prader-Willi syndrome?

Prader-Willi syndrome is a genetic condition due to an abnormality on chromosome 15 and is rarely hereditary. There are 4 different genetic mechanisms causing the defect, which happens at the time of conception, and neither parent is to blame. It can therefore occur in any family. Normally only one child is affected. Siblings almost never pass the condition on to their children. It is important that genetic testing should be performed and that parents should be referred for genetic counselling. This can be arranged through a doctor. Most of the abnormalities described are due to a problem or disorder in the hypothalamus. The hypothalamus is a small gland in the middle of the brain that secretes hormones (growth hormone and sex hormones) and also influences control of appetite.

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How is Prader-Willi syndrome diagnosed?

Awareness of the syndrome has increased and as a result more babies are being diagnosed at birth or shortly thereafter. When a baby presents as floppy after birth the possibility of the syndrome should be verified by specialised genetic testing on a blood sample. Different blood tests are available to confirm the diagnosis.

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What treatment is available?

There is no cure for the disorder. However, recognition and early intervention may improve the prognosis. Treatment targets the specific abnormalities and is managed by a multi-disciplinary team.

- Support with feeding and restricted adequate energy intake to control weight gain under guidance of a dietician. Daily exercise is imperative for weight control and health. Conventional appetite suppressants are of no value, but research is being done on numerous drugs to control appetite.
- Physiotherapy to improve muscle tone, speech and language therapy and occupational therapy for learning difficulties and developmental delay. Specialised education may be incorporated in main stream schools.
- Hormone treatment by an endocrinologist, e.g. growth hormone treatment from early on to improve muscle strength and growth and sex hormone treatment to improve sexual characteristics.
- Treatment for behavioural abnormalities is limited, but daily routines and structure, firm rules and limits, and positive rewards work best for behaviour management. Psychotherapy may be beneficial.
- Qualified professionals should address complications due to obesity, e.g. diabetes, hypertension, breathing problems and depression.

Families have to deal with very strenuous circumstances and therefore parents and other family members should receive counselling to understand and manage the complexities associated with the syndrome.

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What does the future hold for people with Prader-Willi syndrome?

Very few adults with Prader-Willi syndrome are able to lead a fully independent life and most will need continuous support and care throughout their lives. With better management and understanding people with Prader-Willi syndrome are now living well into middle age and beyond and contribute many skills and good qualities to the community in which they live.

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What is the Prader-Willi Syndrome Association of South Africa?

The *Prader-Willi Syndrome Association of South Africa* is a support group and was established by a small group of concerned parents in March 1990.

The main aims of the Association:

- To provide support to parents and caregivers of individuals with Prader-Willi syndrome.
- To promote knowledge and awareness of the syndrome among the public and the medical and paramedical professions.
- To improve the care given to individuals with Prader-Willi syndrome.

We invite everyone involved with individuals with Prader-Willi syndrome, including parents, family members, friends, professionals, caregivers and other interested persons, to become members of the Association.

FOR FURTHER INFORMATION, PLEASE CONTACT:

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The PWSA (SA) is a member of:

International Prader-Willi Syndrome Organisation (IPWSO) www.ipwso.org

South African Inherited Disorders Association (SAIDA) www.saida.org.za

Western Cape Forum for Intellectual Disability (WCFID) www.wcfid.co.za

The South African Association for the Scientific Study of Mental Handicap (SAASSMH)

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