



Prader-Willi Syndrome

<p>What is it?</p>	<p>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.</p>
<p>What are the characteristics of Prader-Willi syndrome?</p>	<p>Numerous characteristics have been described in the literature, but it is important to remember that the clinical picture can vary and changes with age.</p> <p>Main characteristics:</p> <ol style="list-style-type: none"> 1. In the neonatal period: <ul style="list-style-type: none"> • 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: <ul style="list-style-type: none"> • 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 <p>Numerous other features have also been described.</p>
<p>What is the cause of Prader-Willi syndrome?</p>	<p>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.</p>

<p>How is PWS diagnosed?</p>	<p>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.</p>
<p>What treatment is available?</p>	<p>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.</p> <ul style="list-style-type: none"> • 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. <p>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.</p>
<p>What does the future hold for people with Prader-Willi syndrome?</p>	<p>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.</p>
<p>What is the Prader-Willi Syndrome Association of South Africa? PWSA(SA)?</p>	<p>The <i>Prader-Willi Syndrome Association of South Africa</i> is a support group and was established by a small group of concerned parents in March 1990. The main aims of the Association:</p> <ul style="list-style-type: none"> • 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. <p><i>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.</i></p>
<p>FOR FURTHER INFORMATION, PLEASE CONTACT:</p> <p>www.praderwilli.org.za</p>	<p>PWSA (SA) PO Box 2399, Brooklyn Square, Pretoria, 0075, South Africa, Fax: 086 551 5980 Medical advisor: Dr Engela Honey: medic@praderwilli.org.za +27 12 319 2269 Chairperson: Rika du Plooy: chairperson@praderwilli.org.za +27 12 344 0241 Secretary: Wilna: secretary@praderwilli.org.za</p> <p>The PWSA (SA) is a member of: <i>International Prader-Willi Syndrome Organisation (IPWSO) www.ipwso.org</i> <i>South African Inherited Disorders Association (SAIDA) www.saida.org.za</i></p> <p style="text-align: right;"><i>Afrikaans volg</i></p>



Prader-Willi-sindroom

Wat is dit?	Prader-Willi-sindroom is 'n genetiese afwyking. Individue wat hieraan ly, kan aan bepaalde eienskappe uitgeken word soos hipotonie (lae spiertonus), voedingsprobleme en onderontwikkeling van die geslagsdele in die neonatale tydperk en obesiteit, kort liggaamsbou, verstandelike gestremdheid en gedragsprobleme in die latere lewensjare. Die naam is afgelei van die volgende drie dokters wat hierdie toestand in 1956 vir die eerste keer in die mediese literatuur beskryf het: A. Prader, H. Willi en A. Labhart. Die voorkoms is tussen 1 uit 10 000 en 1 uit 15 000 lewende geboortes. Hierdie sindroom kom ewe veel by mans en vroue voor en word by individue van alle rasse aangetref.
Wat is die eienskappe van die Prader-Willi-sindroom?	Verskeie eienskappe word in die literatuur beskryf, maar dit is belangrik om te onthou dat die kliniese beeld van individu tot individu kan verskil en met ouderdom verander. Hoofeienskappe: 1 In die babastadium: <ul style="list-style-type: none">• Hipotonie (lae spiertonus) met 'n sagte of afwesige huilgeluid• Voedingsprobleme wat spesiale hulp in die meeste gevalle verg (bv. gastriese buisvoeding)• Onderontwikkeling van die geslagsdele bv. onafgedaalde testikels• Tipiese gesigstrekke bv. amandelvormige oë, nou voorkop, ens.• Geskiedenis van verminderde fetale bewegings met 'n hoë voorkoms van stuitverlossings 2 Kindertydperk: <ul style="list-style-type: none">• Verhoogde eetlus en buitengewone gewigstoename vanaf ouderdom 2 tot 3 jaar.• Vertraging in die bereiking van vroeë ontwikkelingsmylpale• Gedragsprobleme met woede buie wat meestal gesentreer is om 'n konstante behoefte aan voedsel• Intellektuele inkorting met leerprobleme• Kort liggaamsbou• Afwesigheid van normale seksuele ontwikkeling• Velkrappery• 'n Verskeidenheid ander eienskappe is ook beskryf.
Waardeur word die Prader-Willi-sindroom veroorsaak?	Prader-Willi-sindroom is 'n genetiese toestand wat toegeskryf word aan 'n afwyking op chromosoom 15 en dis selde oorerflik. Vier verskillende meganismes veroorsaak die abnormaliteit. Dit gebeur by bevrugting en nie een van die ouers kan die skuld daarvoor kry nie. Dit kan dus in enige familie voorkom. Gewoonlik word slegs een kind daardeur geraak en ander kinders dra feitlik nooit die toestand aan hul kinders oor nie. Dit is belangrik dat genetiese toetsing gedoen word en ouers kan baat by genetiese raadgewing wat deur enige dokter gereël kan word. Die meeste van die abnormaliteite wat beskryf is, is as gevolg van 'n probleem of afwyking in die hipotalamus. Die hipotalamus is 'n klein kliertjie in die middel van die brein en skei hormone (groeihormone en geslagshormone) af en beheer ook die eetlus.

<p>Hoe word die Prader-Willi-sindroom gediagnoseer?</p>	<p>As gevolg van 'n toename in die bewustheid ten opsigte van die sindroom word al hoe meer babas by geboorte of kort daarna gediagnoseer. As 'n baba baie hipotonies of slap by geboorte voordo, moet die moontlikheid van die sindroom deur gespesialiseerde genetiese toetsing met behulp van 'n bloedmonster geverifieer word. Verskillende bloedtoetse is beskikbaar om die diagnose te bevestig.</p>
<p>Watter behandeling is beskikbaar?</p>	<p>Daar bestaan nie 'n middel om hierdie afwyking te genees nie, maar vroeë diagnose en vroeë intervensie kan die prognose verbeter. Behandeling is gerig op die spesifieke abnormaliteite en word deur 'n multidissiplinêre span bestuur.</p> <ul style="list-style-type: none"> • Bystand met voeding en beheerde energie-inname om gewigstoename te beheer onder leiding van 'n dieetkundige. Daaglikse oefening is noodsaaklik vir gewigsbeheer en gesondheid. Konvensionele eetlusdempers dien geen doel nie, maar navorsing word gedoen op verskeie medikasie om eetlus te demp. • Fisioterapie om spiertonus te verbeter, spraakterapie en arbeidsterapie vir leerprobleme en ontwikkelingsagterstand. Gespesialiseerde onderrig kan in hoofstroomskole geïnkorporeer word. • Hormoonbehandeling deur 'n endokrinoloog bv. vroeë groeihormoonbehandeling om spierkrag en groei te verbeter en later geslagshormoonbehandeling om geslagseienskappe te verbeter. • Behandeling vir gedragsafwykinge is beperk, maar daaglikse roetine en struktuur, streng reëls en beperkings en positiewe belonings werk die beste vir gedragsbestuur. Psigoterapie mag voordelig wees. • Komplikasies as gevolg van obesiteit bv. diabetes, hipertensie, asemhalingsprobleme en depressie moet deur die toepaslike professionele persone behandel word. <p>Gesinne moet met baie moeilike omstandighede saamleef en daarom word berading vir ouers en ander gesinslede aanbeveel om die kompleksiteite wat met die sindroom geassosieer word te begryp en te hanteer.</p>
<p>Wat hou die toekoms in vir mense met die Prader-Willi-sindroom?</p>	<p>Baie min volwassenes met die Prader-Willi-sindroom kan ooit 'n ten volle onafhanklike lewe lei en die meeste van hulle sal voortdurende bystand en versorging gedurende hul leeftyd nodig hê. Met beter bestuur en begrip leef mense met Prader-Willi-sindroom nou tot in hul middeljare en selfs langer en lewer hulle bydraes tot die gemeenskap waarin hulle leef, deur hulle vele vaardighede en goeie eienskappe.</p>
<p>Wat is die Prader-Willi-Sindroomvereniging van Suid-Afrika?</p>	<p>Die <i>Prader-Willi-Sindroomvereniging van Suid-Afrika</i> is 'n ondersteuningsgroep wat deur besorgde ouers in 1990 gestig is.</p> <p>Die hoofdoelstellings van die Vereniging:</p> <ul style="list-style-type: none"> • Om bystand te verleen aan ouers en versorgers van individue met die Prader-Willi-sindroom • Om kennis en bewustheid van die sindroom by die publiek en die mediese en paramediese professies te kweek • Om die versorging van individue met die Prader-Willi-sindroom te verbeter <p><i>Ons nooi almal uit wat betrokke is by 'n persoon met Prader-Willi-sindroom om by die Vereniging aan te sluit. Ouers, familielede, vriende, professionele persone, versorgers en ander belangstellendes is baie welkom.</i></p>
<p>VIR VERDERE INLIGTING KONTAK ASSEBLIEF: www.praderwilli.org.za</p>	<p style="text-align: center;">PWSV (SA) Posbus 2399, Brooklyn Square, Pretoria, 0075, Suid-Afrika Faks: 086 551 5980 Mediese raadgewer: Dr. Engela Honey: medic@praderwilli.org.za +27 12 319 2269 Voorsitter: Rika du Plooy: chairperson@praderwilli.org.za +27 12 344 0241 Sekretaresse: Wilna: secretary@praderwilli.org.za</p> <p style="text-align: center;">Die PWSV (SA) is lid van: <i>International Prader-Willi Syndrome Organisation (IPWSO) www.ipwso.org</i> <i>South African Inherited Disorders Association (SAIDA) www.saida.org.za</i></p>