

Prader-Willi Syndrome - Information Sheet 1

– to be included in the Personal Health Records of people who have a current problem with challenging behaviour

The material in this sheet has been adapted from the Therapeutic Guidelines book 'Management Guidelines for People with Developmental and Intellectual Disabilities' and updated from the 2005 version 'Management Guidelines – Developmental Disability' which can be consulted for more detailed information.

INTRODUCTION

Prader-Willi syndrome is a distinct condition characterised by neurological impairments causing an altered pattern of growth and development with associated hyperphagia (over-eating). This preoccupation with food and accompanying compulsion to eat can cause extreme obesity with premature death. Early intervention with a dietitian and behavioural psychologist to prevent excessive weight gain in childhood is crucial. With appropriate interventions and ongoing support, people with Prader-Willi syndrome are now living into their eighth decade. Most adults live with family or in accommodation with a variable amount of formal support. A holistic approach to care is essential, with clear communication between the patient, family, medical practitioners, other health professionals, educators, and support staff.

AETIOLOGY (CAUSE)

Prader-Willi syndrome is caused by a lack of a functioning paternally-inherited genes in a critical region of the long arm of chromosome 15. Prenatal diagnosis is possible, although risk of recurrence is very low (except when caused by an imprinting anomaly).

INCIDENCE

The condition is mostly sporadic, affecting both males and females and is not associated with any racial, ethnic or socioeconomic group. Estimates of the birth prevalence vary between 1/8,500 to 1/38,000 births.

PRESENTATION

Most children with Prader-Willi syndrome will come to the attention of doctors because of their neonatal hypotonia, developmental delay, and/or obesity.

Presentation in Neonates and Infant

- Neonates may have severe hypotonia (floppiness) with a weak suck, often necessitating nasogastric feeding.
- Severe delay in motor development is common.
- Hypogonadism in boys is common.

Presentation in Children

- The physical phenotype, characterised by short stature, deep-set almond-shaped eyes, narrow forehead and small hands and feet, is usually recognisable during the second year of life.
- There is a delay in sitting and crawling, but the severe hypotonia improves and most children start walking independently between two and three years.
- Young children may present with mild to moderate global developmental delay.
- Learning difficulties associated with a mild intellectual disability are usually obvious by school entry.
- Preoccupation with food usually becomes evident during the pre-school years.
- Food-seeking behaviours are often evident by school age.
- The onset of obesity can be subtle, without obvious change in eating behaviour or calorific intake.
- Short stature usually develops as childhood and adolescent growth spurts may be delayed or absent.
- The behavioural phenotype may become evident during childhood with exacerbations during adolescence.
- Delayed or incomplete puberty is common.

Presentation in Adults

There are probably adults where the condition is not recognised. The diagnosis should be considered in patients with short stature who demonstrate a behavioural phenotype which includes:

- obesity with food-seeking behaviour
- severe delays in adaptive behaviour
- limited speech
- perseverative behaviours

MAKING A DIAGNOSIS

The diagnosis of Prader-Willi syndrome can now be made with a blood test using methylation analysis. This test detects the deletion, uniparental disomy (which are sporadic with no risk of recurrence) and imprinting mechanisms (which can have a recurrence rate of up to 50%). If the test is negative and there is a high index of suspicion referral to a geneticist is indicated to investigate for rare anomalies.

Other diagnostic tests used are FISH to detect deletions and 15 PCR to detect paternal disomy.

OBESITY

There is an extreme risk of obesity due to both the preoccupation with food and lower energy expenditure. Obesity increases the risk of diabetes, coronary artery disease, cor pulmonale, sleep apnoea and premature death. Prevention of obesity is of paramount importance for physical, psychological and social well-being.

Preoccupation with Food

All people with Prader-Willi syndrome have a preoccupation with food along with reduced appetite control. As yet there is no specific drug to counteract this, although there are optimistic reports with SSRI drugs.

Low Energy Expenditure

As there is a lower energy expenditure compared to that of the general population, patients require considerably less kilojoules/ calories than normal. This is partly due to their high ratio of body fat to lean body mass.

Reduced Physical Activity

Hypotonia with reduced muscle bulk and strength persists into adult life and contributes to poor posture and reduced stamina for physical activities.

Prevention and Management of Obesity

Prevention and management of obesity is dependant on a combination of :

- a sensible diet
- an exercise plan
- modification of eating behaviours

The establishment of healthy eating patterns and a low calorie diet prior to the onset of preoccupation with food is essential. The close involvement of a dietitian and behavioural psychologist is necessary to maintain nutritional guidelines throughout a person's life, particularly in settings where there is a high turnover of support staff .

Recommendations

- Children and adolescents should have their growth regularly measured and plotted on growth charts to detect early weight gain, and monitor height velocity.
- Weight ideally should be maintained at less than the ninetieth percentile.
- Adults should be weighed regularly and their body mass index (BMI) maintained at less than twenty five. Monthly to quarterly weighing is usually all that is necessary unless there is rapid weight gain or foraging for food.
- All patients need a dietitian who can assist in developing nutritional guidelines. These need to be individualised and reviewed regularly.
- Dietary guidelines, restricted access to food and promotion of physical activity are essential in all settings; childcare, kindergarten, school, work and home.

Modification of Excessive Eating

The total involvement of family and carers is essential to ensure restricted access to food to achieve dietary control. Specific measures will need to be taken, such as having an inaccessible pantry and supervision of snacks and meals at school and elsewhere, in order to restrict opportunities to over-indulge. Explanation of the eating disorder and diet program to extended family, carers, teachers, employers and others who have contact with the patient is very important. Written information which is individualised, together with general information should also be made available.

Prader-Willi Syndrome - Information Sheet 2

BIOPSYCHOSOCIAL ISSUES

Cognitive Disabilities

Specific learning and/or language disorders may occur with a normal intellect, although the majority of people with Prader-Willi syndrome have an IQ in the mild intellectually disabled range, often with greater than expected difficulties in adaptive behaviour.

Some specific disabilities described include:

- Speech difficulties resulting from both hypotonia and cognitive impairments.
- Short term memory and sequential processing deficits.
- Language processing problems.
- Difficulties in self-reflection and conceptualisation resulting in reduced capacity for self-monitoring.

Recommendations

Early referral to speech pathologist for assessment, advice, therapy and ongoing surveillance is recommended. Some toddlers and preschool children with expressive language delays benefit from Makaton sign language.

Behavioural Phenotype

People with Prader-Willi syndrome are at an increased risk of a cluster of behavioural traits possibly related to specific cognitive impairments. The phenotype includes:

- a preoccupation with food
- other obsessive traits
- mood lability
- impulsiveness
- temper tantrums
- inactivity
- repetitive speech patterns
- a relative weakness in social skills and adaptive behaviour

These difficulties often increase with age. During school years, special education and psychological assistance is often required for difficulties in adaptive behaviour together with environmental adaptations to restrict unsupervised access to food to prevent obesity. Adolescence and early adulthood is frequently a period of emotional lability, defiance, difficulties with socialisation and depression. However there is a natural variation in personal characteristics and abilities to develop coping skills.

Recommendations

- Early referral to psychologist for cognitive and clinical assessment with appropriate on-going advice, support and interventions. Long-term follow up is beneficial for both the patient and their family.
- Referral to a behavioural psychologist for specific behaviour modification intervention may be required.
- Psychiatric and physical illness should be excluded when serious problems arise or when there is a sudden deterioration in behaviour.

Psychiatric Disorders

There is an increased risk in adults with Prader-Willi syndrome of psychiatric disease. Recent reports have noted a relationship between family stress and behavioural symptomatology, with a high risk for obsessive compulsive disorders and an increased vulnerability to psychotic disorders.

Recommendations

After excluding or treating any psychiatric disorder an eclectic approach with a wide range of supports and interventions for behavioural difficulties is required and may include pharmacological, behavioural and/or psychotherapeutic strategies.

HEALTH PROBLEMS

Common Health Problem	Recommendations
Infantile Failure to Thrive	<ul style="list-style-type: none"> Caused by slow and weak suck and swallowing mechanisms. Calorie supplements. Nasogastric feeding may be required.
Short Stature	<ul style="list-style-type: none"> Most young children are at the lower end of the normal height range. Most adults are less than the third percentile in height (males ~1.5m, females ~1.4m). Monitor growth and if velocity is slowing with adequate calorific intake refer to endocrinologist for consideration of growth hormone.
Male Sexual Development	<ul style="list-style-type: none"> Abnormal development of the external genitalia e.g. hypoplastic scrotum, undescended testes, and/or a small penis. Micropenis with awkward micturition. Lack of deepening of the voice. Delayed or incomplete puberty. Surgical repair. A short course of testosterone undecanoate, sustanon 100 may be useful to increase scrotal development and penile size. Consideration of hormone replacement therapy
Female Sexual Development	<ul style="list-style-type: none"> Delayed menarche or scanty, infrequent, irregular menses. Pregnancy has not been reported. Consideration of hormone replacement therapy.
Diabetes	<ul style="list-style-type: none"> Increased risk with obesity. Diet Oral hypoglycemics (metformin).
Osteoporosis	<ul style="list-style-type: none"> Increased risk. Measurement of bone marrow density. Regular exercise.
Scoliosis and Kyphosis	<ul style="list-style-type: none"> Increased risk. Can develop rapidly if obese. Annual reviews, with radiology if obese. If severe or rapid progression, refer to orthopaedic surgeon.
Sleep Disorders	<ul style="list-style-type: none"> Obstructive sleep apnoea. Excessive daytime sleeping. REM sleep disorders. Overnight CPAP. Consideration of neuroleptic medication or referral to neurologist.
High Tolerance to Pain	<ul style="list-style-type: none"> Poses a risk of undetected medical conditions. Beware of late diagnosis, especially of abdominal disease e.g. appendicitis.
Reduced Ability to Vomit	<ul style="list-style-type: none"> Poses risk if poisons are ingested. If ingestion occurs gastric lavage is indicated.
Skin Picking	<ul style="list-style-type: none"> Can lead to persistent sores and infections. Early treatment of infected sores as constant skin picking can lead to cellulitis. SSRI antidepressants may be useful.
Dentition	<ul style="list-style-type: none"> Increased risk of caries due to dental anomalies. Yearly dental checks.
Strabismus and Myopia	<ul style="list-style-type: none"> Common. Patching and/ or surgery for strabismus, with screening for myopia every 2-5 years.

Specialist medical services for intellectual disability and Prader-Willi syndrome exist in major teaching hospitals and clinical genetic services in most states.

SUPPORT ASSOCIATIONS

Prader-Willi associations are a valuable source of information and support for parents, professionals and other interested persons. They have available for loan and distribution a wide range of written materials including specific topics for parents, siblings, teachers, community groups and allied health professionals.

Prader-Willi Association of Australia
Victoria/Tasmania

(02) 4946 9001 <http://www.pws.org.au>
(03) 9735 5199 <http://www.pws.asn.au>