Health Checklist

Endocrine / Hypothalamic Issues – GHD, hypogonadism, premature adrenarche (14-30%), hypothyroidism (14-25%) - often CE-H, osteoporosis risk (check BMD, calcium and vit D), possible central adrenal insufficiency under stress. Temperature regulation abnormalities (risk of hypothermia / hyperthermia) and high pain threshold - can mask injury, infection, illness.

Obesity Issues – Refer to dietitian. Compared to primary obesity - lower lean mass / increased adiposity, reduced REE / metabolic rate, early onset, primarily central and unusual fat distribution. Increased risks for: hypertension (usually pulmonary), hypoventilation, oedema, cellulitis, thrombosis and possibly T2DM. Risk factors for T2DM not fully understood but 7-24% prevalence, early onset – monitor glucose homeostasis.

Respiratory & Sleep Issues – Silent aspiration and chronic respiratory infections common in infants. High rate of apnoea (CSA infants, OSA older children / adults). Weak chest muscles, hypoventilation with obesity, altered ventilatory control responses to hypoxemia and hypercapnia, increased risk for pulmonary embolism. Common sleep problems - altered sleep architecture, EDS and sometimes narcolepsy. Sleep study advised for severe hypotonia, severe obesity, EDS, snoring, respiratory problems or starting GHT.

Gastrointestinal Issues – High prevalence: subclinical dysphagia, GERD, slow gastric motility / gastroparesis, chronic constipation. May require dietary changes.

Hyperphagia Alerts – Risk of serious gastric illness, necrosis or rupture. Poor vomit reflex – of concern following food poisoning or binge. Evaluate any abdominal discomfort, bloating, distention, vomiting or odorous belching (pain & fever may be absent.) Swallow dysfunction and voracious eating habits increase choking risk.

Orthopaedic Issues – Flat feet, ankle overpronation, joint laxity, osteoporosis. Infant hip dysplasia (8-30%) – new thinking unsupportive of aggressive treatment. Kyphosis and scoliosis (60-70%) can remain well hidden in PWS – annual radiographic screening advised from sitting to 4yrs, then thorough spine exams, close observation from 10yrs.

Vision Checks – Strabismus and astigmatism common; amblyopia and deficiency in stereopsis may occur. Refer to ophthalmologist. Vision checks – glasses sometimes required.

Dental Checks – GERD and viscous saliva with low salivary production increase cavity risk. Drinking water regularly, saliva stimulation products and good oral hygiene help.

Skin Picking – Very common, open sores vulnerable to infection.

Accelerated Ageing – Biologically proven. Health checks and early specialist screening warranted. Increased cardiovascular risk - CAD reported at young ages.

Medications, Anaesthesia – Prolonged or unusual responses, especially to psychiatric, pain, sedation or antihistamine meds. Drug interaction risks, especially after surgery.

How we can help ...

THE PWSA(NZ) SUPPORTS PEOPLE LIVING WITH PWS, THEIR FAMILIES AND CAREGIVERS

ADVOCACY

- Providing advocacy services on behalf of people living with PWS and campaigning on relevant issues.

EDUCATION

- Accessible information via our website and our range of free publications and resources
- Training offered to schools and residential care providers
- Increasing awareness and understanding of PWS
- Circulating research to medical professionals as needed
- Keeping members informed of the latest news, ideas and research through our newsletters
- Links to the international PWS community

SUPPORT

- Offering information, diagnosis and crisis support to people living with PWS, parents, whānau and caregivers
- Facilitating connection via our parent support network
- Hosting events, family meetings and support weekends.

PLEASE CONTACT US

In an emergency, please see our 'Medical Alerts' booklet. To learn more about PWS and view further professional resources, please visit our website.

Additional copies of this pamphlet can be ordered via our website or by contacting us as below.

Free PWSA(NZ) membership is available to all. Most services offered to people living with PWS and their families are provided free of charge.

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Prader-Willi Syndrome Association (NZ) Incorporated
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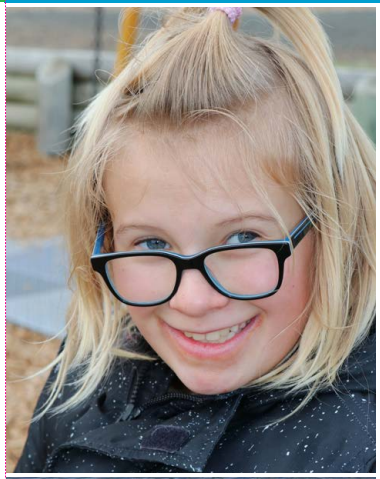
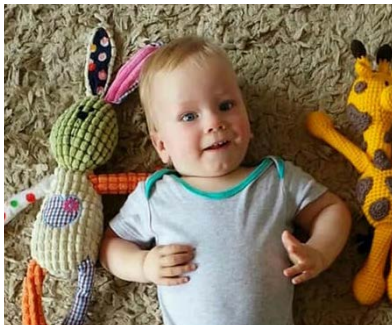
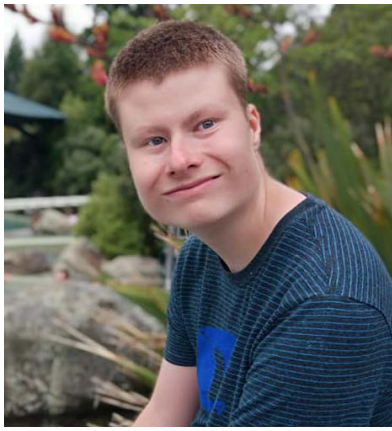
An introduction to Prader-Willi Syndrome for Healthcare Providers




ADVOCACY | EDUCATION | SUPPORT

PWS PRADER-WILLI SYNDROME ASSOCIATION NEW ZEALAND

Prader-Willi syndrome is a complex neurodevelopmental disorder resulting from an abnormality on the 15th chromosome. PWS randomly occurs in approximately 1 in 16,000 births, equally affecting males, females and all ethnicities.

Symptoms include impacts on cognition, emotional regulation, growth, muscle development, metabolism and appetite, with many of these attributed to hypothalamic dysfunction.

CHARACTERISTICS

- failure to thrive
- hypotonia
- slow growth / short stature

} GHT improves body composition and normalises height

- distinct facial features, hypopigmentation (more likely in DEL)
- sleep disorders and excessive sleepiness (EDS)
- developmental delay, possible ongoing motor planning, balance, coordination or articulation difficulties
- hyperphagia - increased hunger, preoccupation with food, enhanced sense of food reward
- specific learning disabilities, often mild intellectual disability
- behavioural challenges linked to anxiety, rigidity, self-regulation
- hypogonadism causing delayed and/or incomplete puberty, usually infertility.

The 2 most common causes of PWS are paternal deletion (DEL) at 15q11-q13 and maternal uniparental disomy (UPD). Less frequently, imprinting defects (ID) or rare translocations occur. Subtle differences exist between genetic subtypes with no feature found exclusively in one group. Each person with PWS will be affected differently, and as PWS is a spectrum disorder, the presentation and severity of each symptom can vary and may change over time.

DIAGNOSIS

PWS is usually diagnosed in infancy today, enabling early interventions, dietary plans and treatments to be implemented, greatly improving prognosis. To avoid cases being missed, the current recommendation is to test all infants with unexplained hypotonia and poor suck. Other diagnostic criteria for infants include failure to thrive and genital hypoplasia. Male cryptorchidism is common. Subtle, yet distinct facial features may also be apparent: a narrowing at the temples, a small down-turned mouth with thin upper lip and almond-shaped eyes. Mothers will typically experience reduced fetal movements during pregnancy with other common symptoms, such as polyhydramnios and malpresentation. The diagnosis of PWS in older children requires a history of hypotonia and poor suck, as well as: global developmental delay (0-6yrs), excessive appetite (6-12yrs), cognitive impairment, hypogonadism and typical PWS behaviours (13yrs+). DNA methylation analysis will diagnose PWS by all genetic mechanisms.

PRESENTATION, DEVELOPMENT & MANAGEMENT

Assisted feeding is typically required for a few weeks to several months. Contrary to traditional thinking, some babies have been able to breast feed with persistent offering of the breast before a tube feed, although this takes longer to establish. Global developmental delay occurs, but attainment of physical milestones can be accelerated or normalised by growth hormone treatment. Motor and speech problems might be ongoing.

Weight gain without excess calories can begin from 18mths-3yrs indicating the need for calorie restriction with a reduced carbohydrate diet. Food has intensified significance from around 4.5yrs, with onset of hyperphagia from around 8yrs. With an altered metabolism and high risk of food seeking behaviour, establishing **'Food Security'** can prevent rapid weight gain and reduce food related anxiety: **No Doubt + No Hope (or chance) = No Disappointment** (Pittsburgh Partnership, USA).

Most people living with PWS will have mild to moderate intellectual disabilities alongside typical learning difficulties, such as auditory processing disorder, memory problems, dyscalculia and impaired executive functioning. Rigidity in thinking, sensory processing issues, anxiety and poor social skills are very common. The cognitive profile has overlaps with ASD, particularly in the areas of compulsivity and insistence on sameness - a few children receive co-occurring diagnoses. ADHD (inattention /impulsivity) is also common. In adolescence and early adulthood, challenging behaviours typically intensify and additional supports are required. Proactive rather than reactive behaviour management strategies that minimise anxiety work well: **The 4Cs = Be Calm, Consistent, Clear + avoid Confrontation.**

GROWTH HORMONE THERAPY

Described as "a potent force for counteracting the clinical course of obesity in PWS," GHT should be considered for all patients with PWS, both children and adults. At time of publication, GHT is only funded from 6mths until end of growth (unless patient has severe GHD), but some families privately fund treatment before 6mths or into adulthood (ask us about the possibility of assistance for adults.) Studies have demonstrated that in addition to building strength, GHT has benefits for motor development, speech, cognition, plus mental health and quality of life in adulthood.

Research has observed that benefits increase with earlier treatment, so it is vital to make the necessary referrals as soon as possible following diagnosis. At end of growth, GHT helps maintain body composition and exercise capacity. Detrimental effects occur following cessation of GHT and symptoms of adult GHD can develop. Reducing dosage gradually to an appropriate maintenance dose can prevent rapid increases in BMI (VAT and LDL) and sudden behavioural change.

OTHER THERAPIES & TREATMENTS

Physio and Occupational Therapies

Hypotonia causes lifelong problems with decreased strength and endurance, hypermobile joints, articulation, and poor posture. A high prevalence of scoliosis (60-70%) necessitates programmes that target the core and upper body. Orthotic supports may be beneficial. GHT improves body composition in conjunction with PT and dietary control, but exercise needs to be a regular part of daily life. Support is needed for motor skill development (fine motor better than gross). Visual-motor difficulties and symptoms of dyspraxia are typical. Sensory and vestibular input via sensory integration therapy is helpful for developing balance and motor planning ability.

Feeding / SLT Therapy

A high rate of dysphagia increases risk for reflux and silent aspiration in infants - videofluoroscopic assessment (VFSS) is recommended and specialist advice on feeding techniques, if needed. Oral-motor therapy in infancy can 'awaken' muscles ready for speech. Speech delay is typical as hypotonia causes symptoms associated with dysarthria. Early signing can help bridge communication gaps. Ongoing oral-motor and articulation difficulties are common, possibly due to hypotonia, motor deficits, oral structure and saliva abnormalities, or poor phonological skills. Apraxia of speech may occur in up to 50% and PROMPT therapy has proven useful when expressive language is sufficiently developed.

Sex Hormone Treatments

TRT and ET/EPT are recommended for promoting and maintaining pubertal development, retaining muscle mass, general wellbeing and bone health (BMD). Optimal starting age is 14.5-16yrs boys / 13-16yrs girls at low dose with gradual increases and treatment continued through adulthood. Risk of TRT mood instability can be ameliorated by reducing IM injection intervals or administering using alternative forms.

Psychiatric Treatments

Anxiety, skin picking, and ritualistic or 'needing to know/ask/tell' behaviours are typical, but OC symptoms usually differ to classic OCD. There is increased risk for mood disorders or psychotic illness, particularly in adolescence / early adulthood and in UPD. Depression can easily be overlooked and bipolar disorder may present differently (true mania is rare). Atypical side effects to medications due to metabolism differences or neurosensitivity include greater response to SSRIs and mood activation at typical dose. Advice is to start low, go slow.

Children should see their primary clinician at least 6 monthly, ideally as part of a multidisciplinary team. Annual health checks with blood biochemistry are advised for adults.