Syndrome Specific Medical health check guide - Prader-Willi Syndrome (PWS)

Introduction
Prader-Willi Syndrome (PWS) is the most common genetic form of obesity and is usually characterised by voracious overeating, hypotonia and morbid obesity. In the UK, the prevalence of PWS is estimated to be one in 15,000 to 25,000 and is seen equally in all ethnic groups. Prader-Willi syndrome is caused by the loss of function of genes in a particular region of chromosome 15.

People with PWS face complex health issues with a relative risk of death much higher than other people with ID. Early death as a result of morbid obesity is not uncommon, however, with restricted access to food, the person can live well into middle age and potentially beyond 50 years of age. Somatropin (recombinant human growth hormone) is recommended as a treatment option for children with growth failure associated with PWS by NICE.

History
Characteristics of PWS vary considerably between individuals and may include: Hyperphagia (overeating) and consumption of life-threatening amounts of food. There can also be ingestion of inappropriate items such as out-of-date food or food from waste bins. Gastroparesis and constipation may occur after binge and bowel necrosis and stomach rupture have been reported. The drive to eat overwhelms rational decision-making.

History of rare vomiting or lack of vomiting reflex, probably due to low muscle tone. If the person has abdominal pain or distension there may be life-threatening bowel obstruction or necrosis. Delayed stomach emptying is common with people with PWS and can be life-threatening.

Absence of fever even when seriously ill, and may run dramatically below-normal temperatures at times. Even slight temperature elevations should be considered a warning sign to check for further symptoms.

Unreported injuries or undetected bone fractures, due to high pain threshold. In addition, osteoporosis is more common because of the hypotonia and lack of sex hormone.
Skin picking, which may seriously worsen at this (WHICH?) age, and can be exacerbated by anxiety. It is important that carers and paid staff monitor for infections. Oedema and leg swelling is common.

Excessive weight combined with the hypotonia of PWS leading to serious respiratory problems. Obstructive sleep apnoea and respiratory failure are the most common problems for obese people with PWS. Congestive heart failure and pulmonary hypertension may also occur at this age due to significant obesity.

Drinking excessive amounts of fluid, leading to potentially fatal hyponatraemia and hypokalaemia.

Bruises, but because of the high pain threshold often unable to say how they came by the bruise.

Hypogonadism. In the majority of cases, full sexual development does not occur in either men or women with PWS. Women may not experience the onset of periods and breast development may be slow. Men’s voice may not break and facial hair can be very scanty. Sex hormone treatment will help with these issues – specialist input from an endocrinologist should be sought. Most patients will be infertile but sex hormone replacement will be required throughout life for bone health.

Mental health problems, which can include: depression, severe anxiety, lethargy, visual and auditory hallucinations, and acute psychotic episodes.

**Examination**

- Weight, height and BMI on a regular basis
- Blood pressure

**1. Musculoskeletal**

- Spine for Scoliosis and Kyphosis
- Osteoporosis (in both males and females, at young age)
- Skin for bruising, picking, oedema and infection

**2. Blood tests**

- Annual blood tests:
  - TFTs (Hypothyroidism)
  - Glucose and HBA1c (Up to 25% of adults with PWS have Type 2 diabetes. In children and adolescents, about 4% have impaired glucose tolerance)
• Lipid profile
• Vitamin D (Deficiency)

Resources

PWS Association, information for GPs: https://www.pwsa.co.uk/pdfs/information-for-gps/