

Information on people with Prader-Willi Syndrome (PWS) for Health Care Professionals

Summary: PWS is a genetic disorder characterised by a dysfunction relating to the hypothalamus which causes the features associated with PWS namely:

- Hyperphagia -excessive appetite from about the age of 2 but sometimes later
- Hypotonia –weak muscle tone which can be improved with exercise
- Hypogonadism –immature sexual development in males and females
- Typically short stature (less so in tall families), small hands and feet
- Learning disability ranging from mild to severe
- Emotional problems including temper outbursts and stubbornness

Due to the hypothalamic dysfunction, there is poor body temperature regulation, high pain threshold and, with the additional lack of vomiting, a person with PWS can have a serious medical problem which may remain overlooked.

Features of PWS requiring particular attention

Vomiting/Abdominal Pain

People with PWS do not normally vomit. Where pain, flatulence, tummy distension and vomiting are present this could be life threatening where emergency surgery is required. It may also be a sign that a large amount of food has been eaten. In either case death can occur in hours although this is rare.



Temperature

An individual with PWS may not present with a fever even when seriously ill and may run dramatically below normal temperature at times. Even slight temperatures should be considered a warning sign when looking at other symptoms and signs of ill health.



Food Foraging

If unsupervised, some with PWS may consume life threatening amounts of food including out of date or frozen food. Abdominal pains and or vomiting may be a sign that a person is seriously ill. Stomach rupture is possible. Diarrhoea or significant fluid retention can also be reasons for concern.



Skin Lesions

A common feature of PWS is skin picking, occasionally severe. Those with open sores should be monitored for signs of infection. Cellulitis may be more common especially if a person is overweight.



High Pain Threshold & Bruising

People with PWS often have decreased sensitivity to pain. They may also bruise easily, common in PWS with no obvious explanation. Reported injuries must be assessed and closely observed for more serious problems.



Where pain is not reported, observe for other signs of injury e.g. bruising, swelling or bone fractures. Do not expect the person with PWS to necessarily complain of pain whether ill or injured.

Anaesthesia

There is nothing inherent in PWS which gives cause for concern with the administration of anaesthesia. However, individual health problems related to PWS should be taken into account including:

- Obesity (complications caused by obstructive sleep apnoea, pulmonary hypertension, altered blood or oxygen or blood carbon dioxide levels and significant oedema)
- High pain threshold (see above)
- Temperature instability-parent or carer should be asked for information about patient's usual temperature
- Thick saliva-may complicate airway management
- Food seeking behaviours-the person may have eaten food even if they say they have not. Unless a parent or carer can verify this, it should be assumed that food is in the stomach
- Hypotonia which may cause difficulties in ability to cough and clear airways
- Excessive post operative drowsiness in some individuals



Mental Health Problems

Some teenagers and adults with PWS may also experience mental health problems. These can include depression, lethargy, hallucinations and hearing voices and acute psychotic episodes, often with a rapid onset.

Respiratory Problems Obstructive Sleep Apnoea

Excessive weight together with poor muscle tone (common in PWS) can lead to serious respiratory problems. Sleep apnoea is common but can also occur in those not obese.



Excessive Fluid Intake

There have been reports of people with PWS drinking excessive amounts of fluid leading to potentially fatal low sodium and potassium levels.



Obesity Related Problems

High blood pressure, diabetes, congestive heart failure and respiratory failure are the most common problems for the person with PWS who is overweight.



Further information: PWSA (UK) 125a London Rd, Derby, DE1 2QQ Tel: 01332 365676 Fax: 01332 360401 email admin@pwsa.co.uk or visit www.pwsa.co.uk or www.pwsausa.org