

Prader-Willi Syndrome (PWS) – Information for Emergency Department (ED) and other hospital staff

Prader-Willi Syndrome (PWS) is a rare genetic condition which is present from birth. Caused by an abnormality on chromosome 15, it affects the functioning of the hypothalamus and other aspects of the brain and muscles. Main characteristics of the syndrome are:

- **Hyperphagia** – beginning around 2-4 years
- **Hypotonia** – from birth
- **Hypogonadism**
- **Short stature, compared with other family members** (less evident if receiving growth hormone treatment)
- **Delay in and difficulties with emotional and social development**
- **Developmental delay /learning disabilities ranging from severe to borderline.** A minority of people with PWS do not have a general learning disability. Most have specific difficulties eg - slow processing of verbal information etc
- **Challenging behaviours**, (there is a PWS behavioural phenotype), whether or not the person has a learning disability.
- A significant number of individuals with PWS also have **Autistic Spectrum Disorder** or some features thereof.

Unusual features and potential risk factors in PWS

A high pain threshold, poor temperature regulation and lack of vomiting associated with hypothalamic dysfunction can mean that a serious medical problem may present in an unusual way and so could be overlooked.

Abnormal body composition affecting medication

Individuals with PWS have reduced lean tissue and increased adiposity. This may affect dosages of medication, which may need to be less than usual. Psychotropic medications must always be started at very low doses. Medications that have antidiuretic effects may cause water intoxication.

Body temperature abnormalities

An individual with PWS may be unaware when they are too hot or too cold, or eat or drink very hot food or liquids. Hyper and hypothermia have been reported. Hyperthermia may occur during minor illness and after anaesthesia. Fever may be absent despite serious infection. In cases of hypothermia, the patient may not report feeling cold. Baseline temperature may be below 98.6°F/37°C.

High pain threshold

The majority of people with PWS have a high pain threshold and hence underreport pain. This may mask fractures, acute abdominal conditions, serious infections and internal injuries. Individuals may have difficulties localising pain.

Lack of vomiting

Vomiting rarely occurs in PWS but some children with PWS do vomit with minor illnesses. The absence of vomiting cannot therefore be relied upon as reassuring when assessing illness. However, the presence of vomiting, particularly when the individual has a history of never or rarely vomiting, may signal a very serious or life-threatening illness (see below). Emetics may be ineffective.

Severe gastric distension with ischaemia

This is a life-threatening situation that may result in stomach rupture. The person with PWS may present with abdominal distension, pain and/or vomiting but may only complain of mild abdominal discomfort. This gastric inflammation with necrosis most often follows a recent binge episode by an individual with PWS whose weight is generally under control.

Gastroparesis and slow gastrointestinal transit

Many individuals with PWS also have gastroparesis, which can become dangerous if overeating occurs or has occurred. Gastroparesis may be caused by viral illnesses and can require decompression with a nasogastric tube or treatment with IV fluids. Most individuals have slow gastrointestinal transit and illness and dehydration can make this worse and result in pseudo obstruction; vomiting is unusual even if this happens.

Loss of appetite

People with PWS will usually eat even when ill, due to hyperphagia. Loss of appetite should be viewed as possibly symptomatic of a serious disorder. However, some people with PWS will refuse food as a controlling behaviour.

Skin lesions – picking and bruises

Many individuals with PWS pick at small wounds or spots, intensifying and increasing the wound, and thus the potential for infection. This can complicate the healing of IV sites and incisional wounds. It is common for individuals with PWS to have sores caused by their own skin-picking. They may bruise easily. Appearances may wrongly lead to suspicion of abuse.

Tendency to psychosis

There have been a few anecdotal reports of teenagers and adults with PWS experiencing psychotic episodes during hospital stays. If parents and carers report worsening differences in behaviour, seek psychiatric help.

See next pages for general care and treatment while an individual with PWS is in hospital

General care while an individual with PWS is in hospital

Informed consent (adolescents and adults)

Anxiety is common in both adults and adolescents with PWS and individuals benefit from receiving clear information about what is going to happen. Even those with higher cognitive functioning may become anxious or confused about proposed treatments (e.g. insertion of IV lines). This might trigger challenging behaviour. They will require a calm, patient and understanding approach and support from a learning disability liaison nurse if possible.

Lower calorie diet

Severe obesity can be caused by the combination of hyperphagia and hypotonia. However, in individuals whose access to food has been well controlled, obesity may not be evident. In **all cases**, however, energy intake should be around two thirds of the average for the individual's age group, whilst maintaining a balanced, nutritious diet. The input of a dietitian may be required. Their usual routines around food should be established and followed.

Restrict access to food

Hyperphagia causes food-seeking and food-stealing behaviour. Ensure that the individual with PWS is not left alone with food or drinks trolley or drinks vending machines, nor has access to other patients' food and plates. Be aware that an individual may say that they have not had a meal or snack when they have already eaten.

Comprehension and language – receptive and expressive ability

Many people with PWS can give the impression that they understand everything said to them. This is not always the case. Others may have articulation problems or dyspraxia and hence be difficult to understand. Children and adults may interpret information literally. Keep instructions clear and simple. Use visual aids to help with both comprehension and communication. Give them plenty of time to process information and respond.

Time frames

Be very clear about time frames. If you say you will be back in a few minutes, make sure you do so. Not sticking to times given can result in a rapid increase in anxiety and escalating challenging behaviour. It is helpful to be very clear about meal times and try to keep to them.

Parental support

Individuals with PWS require structure to their lives and can find unexpected events or change difficult. The hospital environment can be a very challenging time for them. Parental or carer support and access may be needed much more frequently than usual, especially for adults in wards with restricted visiting times.

For children, take parental advice about their child's routines and expectations particularly about food. Small changes in expectations around food can have significant consequences. Take parents' lead on ways of reducing anxiety and responding to any behavioural difficulties.

Behavioural and emotional outbursts

Children and adults with PWS are prone to emotional lability and outbursts. The reasons for an outburst are not always clear but may include hunger, tiredness, uncertainty, unexpected events, conflict etc. Parent or carer guidance can help to anticipate triggers and minimise difficulties.

Treatment

Anaesthesia

Anaesthesia recommendations for patients suffering from Prader-Willi syndrome
www.orpha.net/data/patho/Pro/en/Prader_Willi_EN.pdf

Cortisol levels

Individuals with PWS are probably more likely than the rest of the population to have cortisol deficiency. If there is clinical suspicion of cortisol deficiency during acute illness, immediately take a blood sample to check serum cortisol level. This can then be used to help with diagnosis even if you need to start hydrocortisone as an emergency. Please discuss results with your local endocrinology team.

Medication

People with PWS may have abnormal reactions to standard doses of medication (see Abnormal body composition - pg 1)

Patients receiving growth hormone replacement

Growth hormone (GH) replacement is often used in patients with PWS. If felt necessary, it can be temporarily stopped during an acute illness without any likely acute adverse consequences. Patients and families may however wish to continue GH to help maintain routine. Please discuss with your local endocrinology team if any questions or concerns.

Oxygen levels

Oxygen levels may be normally lower than usual. Obese patients who have been chronically hypoxic may not tolerate fully corrective use of oxygen and are likely to start retaining CO₂.

Additional information

Guidelines for postoperative monitoring of patients with Prader-Willi Syndrome

www.pwsausa.org/wp-content/uploads/2016/01/Postoperative-Monitoring-of-Patients-in-PWS-MA1050.pdf

Guidelines for postoperative monitoring of paediatric patients with Prader-Willi Syndrome

<https://www.pwsausa.org/wp-content/uploads/2016/01/Postoperative-Monitoring-of-Pediatric-Patients-with-PWS-w-logo.pdf>

Obesity in PWS

<http://pwsausa.org/wp-content/uploads/2015/10/ObesityStillKills.pdf>

Clinical Presentation of Obesity Hypoventilation and Right Heart Failure in Prader-Willi Syndrome

<http://pwsausa.org/wp-content/uploads/2015/10/Clinical-Presentation-of-Obesity-Hypoventilation-and-Right-Heart-Failure-in-PWS.pdf>

Psychiatric alert for Prader-Willi syndrome

www.pittsburghpartnership.com/handouts/Psychiatric%20Alert%202009.pdf

Medical alerts in several languages + library of medical articles

<https://www.ipwso.org/medical-professionals>

Websites with extensive information about PWS

PWSA UK <https://www.pwsa.co.uk/>

International PWS Organisation <https://www.ipwso.org/>

PWSA USA www.pwsausa.org

PWSA UK is in contact with medical specialists with an expertise in PWS. Please do not hesitate to contact us for further information at the address below.

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