PWS PRADER-WILLI SYNDROME ASSOCIATION NEW ZEALAND

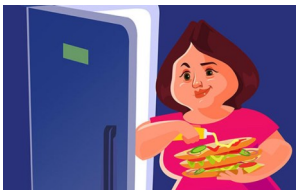
Prader-Willi Syndrome (PWS)
Medical Alerts

PWS is a complex neurodevelopmental disorder resulting from an abnormality at chromosome 15. It causes hypothalamic dysfunction and features such as hypotonia (low muscle tone), hyperphagia, altered metabolism, hypogonadism, learning disabilities, anxiety and impaired self-regulation.

Vomiting / Abdominal Distention or Pain

Urgently evaluate abdominal bloating, distention, discomfort or pain, appetite loss, and/or vomiting (less common in PWS), extra flatus or odorous belching. Life-threatening inflammation, necrosis or rupture can occur, especially after suspected binge eating. Slow gastric motility, constipation or obstruction are common. Signs of infection may be absent (fever or localised pain). Vomiting may be weak due to hypotonia, which creates a choking or aspiration risk and the recovery position should be adopted. A lack of vomiting is also of concern during gastric illness or if poisons have been consumed.

Hyperphagia (Excessive Eating)



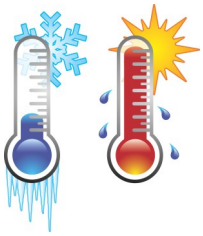
Dangerous volumes of food may have been consumed, or unhygienic, frozen, raw foods, inedible items or poisons.

Plus, excessive fluid intake may cause electrolyte imbalances and cause water intoxication (potentially fatal low sodium.)

High Pain Threshold

Some people with PWS have decreased sensitivity to pain which can mask injuries or illness. Pain may not be felt until severe. Note any subtle changes in behaviour. A person may also be unable to localise or describe their pain well.

Body Temperature Irregularity



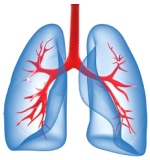
Normal body temp may be altered. Temperature instability may cause a person to not present with a fever when seriously ill, or temperature may be higher than expected (or idiopathic). Slight changes in temperature should be evaluated. Hyper and hypothermia can occur (impaired peripheral and central thermoregulation.)

Anaesthesia

Possibility of prolonged and exaggerated response to sedatives or pain relief. Unless confirmed otherwise, assume a patient may have eaten before surgery. Monitor closely during and post-op considering possible health issues related to PWS:

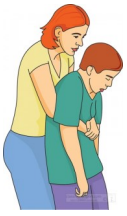
- thick saliva & narrowed oropharyngeal space (airway management)
- hypotonia
- CSA
- decreased pulmonary reserve due to chest wall deformity / scoliosis
- obesity (pulmonary hypertension, altered O2/CO2 levels, OSA, oedema)
- increased risk of PE
- temperature instability
- increased risk of gastric aspiration and choking
- slow GI function recovery
- high pain threshold
- primary myocardial involvement
- patient may have glucose intolerance, diabetes, hypothyroidism or CAI.

Respiratory Concerns



Possible complications from hypotonia and potential apnoea (CSA/OSA). More frequent problems with young age, narrow airway, or morbid obesity. Monitor when unwell. Silent aspiration and hypoventilation are common. Altered ventilatory control responses. PE risk.

Swallowing and Choking Risk



Dysphagia (swallow dysfunction) is typical. Choking risk due to voracious eating habits, oral/motor coordination, poor gag reflex and hypotonia. Reduced and thick saliva flow, so maintain hydration. Supervise. Know first aid.

Skin Sores & Infection

Skin picking is common, sometimes severe. Monitor open sores for infection and cellulitis.

Diabetes Mellitus

Increased risk with some meds and after puberty (avg. 20yrs). Observe for signs of high blood sugar.

Mental Health Problems

High prevalence, particularly in adolescence and early adulthood, often with rapid onset. Can include acute psychosis. Unusual reactions to standard doses of medications.