

## **Important Medical Facts about Prader-Willi Syndrome (PWS)**

(Consensus Document approved by the Clinical & Scientific Advisory Board for IPWSO, January 2022)



**Comprehension and language:** Patients with PWS might give the impression that they understand everything said to them even when they do not. Keep instructions clear and simple, repeat important points and allow time for them to comprehend. Self-reporting of health concerns may be underemphasized or exaggerated, and some have difficulty in articulating health needs. Caregivers accompanying the patient will help by providing or interpreting information.

**Mental health:** People with PWS may have a history of problem behaviours, such as emotional outbursts, and may also develop mental illness which can be of sudden onset and be associated with abnormal mental beliefs and experiences and confusion. Both physical and psychiatric evaluation may be indicated to arrive at a diagnosis under such circumstances.

**Temperature abnormalities:** Hyper- and hypothermia can be idiopathic, as well as occur during major and minor illnesses and in procedures requiring anaesthesia. Fever may be absent despite serious infection. Malignant hyperthermia is not a known risk of anaesthesia for patients with PWS.

**Pain insensitivity:** Lack of typical pain perception is common and may mask the presence of infection, injury, or fractures. Patients may not complain of pain until the condition is severe, and they may have difficulty localizing pain. Any complaint of pain should be taken seriously.

**Hyperphagia:** Patients must be constantly supervised to prevent access to food. They might obtain unguarded food, which can lead to rapid ingestion and fatal choking. Foraging for discarded and spoiled food has occurred. Dysphagia (dysfunctional swallowing) is universal and may contribute to choking and silent aspiration.

**Vomiting:** Vomiting rarely occurs. The presence of vomiting, and/or loss of appetite, may signal a life-threatening illness and may warrant immediate treatment. When induction of vomiting is considered, such as after hyperphagia or the ingestion of uncooked or spoiled food items, emetics are typically ineffective, and repeated doses may cause toxicity. Instead, a nasogastric tube should be used for decompression of the stomach.

**Severe gastric illness:** Abdominal distention, bloating, pain, and vomiting may be signs of life-threatening gastric inflammation or necrosis. There may be a general feeling of feeling ill. Patients with these symptoms should be evaluated by a medical professional and may need to be hospitalized. Radiographic imaging and possibly emergency surgery may be required. Anti-diarrheal medications can cause severe colonic distension, necrosis and rupture and should be avoided.

**Water intoxication:** Water intoxication has occurred either with use of medications with antidiuretic effects or solely from excess fluid intake. A low plasma sodium can help to diagnose this.

**Respiratory concerns:** Patients might be at increased risk of respiratory problems. Hypotonia, weak chest muscles, and sleep apnoea (both obstructive and central) are among possible complicating factors. Significant snoring needs evaluation for obstructive sleep apnoea.

**Adverse reactions to medication:** People with PWS may have unusual reactions to standard dosages of medications. Use extreme caution in giving medications, especially those that may cause sedation; prolonged and exaggerated responses have been reported. The presence of obesity may also affect appropriate dosing.

**Skin lesions and bruising:** Skin picking is commonly seen in PWS, and open sores caused by skin picking may be apparent, sometimes leading to serious infections. Individuals with PWS also tend to bruise easily. Appearance of such wounds and bruises may erroneously lead to suspicion of physical abuse.

To read the full Medical Alerts Booklet for people with PWS please refer to the latest edition from PWSA | USA by scanning this QR code or visiting this link:

<https://www.pwsausa.org/wp-content/uploads/2022/04/MedicalAlertsBooklet-GIChart-2022.pdf>



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