

Important Medical Facts about Prader-Willi Syndrome (PWS)

(Approved by the Clinical & Scientific Advisory Board for IPWSO, January 2022; revised August 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 comprehension. Self-reporting of health concerns may be underemphasized or exaggerated because of challenges articulating health needs. Caregivers accompanying the patient can provide and interpret information.

Mental Health: People with PWS may have a history of problem behaviours, such as emotional outbursts, and may also develop sudden onset mental illness associated with abnormal mental beliefs, experiences, and confusion. Both physical and psychiatric evaluation may be required for thorough assessment.

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: Diminished 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 excess 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 needing immediate treatment. Emetics are typically ineffective for induction of vomiting after ingestion of uncooked/spoiled food items, with potential toxicity from repeated administration. Instead, a nasogastric tube should be used for gastric decompression.

Acute Digestive Emergencies: Abdominal distention, bloating, pain, lethargy, loss of appetite, and vomiting may be signs of life-threatening gastric inflammation or necrosis. Patients with these symptoms should be urgently evaluated by a medical professional and may require hospitalization. Radiographic imaging and possibly emergency surgery may be required. Recommend against use of anti-diarrheal medications due to severe colonic distension, necrosis and rupture. Rectal picking can result in rectal bleeding and iron-deficiency anaemia.

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 Medications: 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 (including rectal picking) is commonly seen in PWS, and open sores caused by skin picking may be apparent, sometimes leading to serious infections including cellulitis and osteomyelitis. 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/resources/medical-issues-a-z/>



Acknowledgement: *We thank Consensus Support Services for an initial draft of this document which has been edited by IPWSO.*