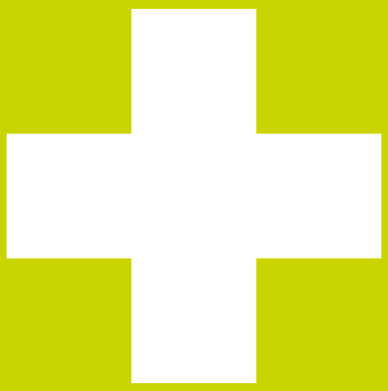
BE ALERT

PRADER-WILLI SYNDROME (PWS)



Emergency Procedures

Medical Alerts and Complications in this rare, genetic condition

An Essential Guide for Medical and Healthcare Professionals

and the Families of Individuals with PWS

consensus...
gretton PWS services
Supporting opportunity, choice and success

Emergency Procedures, General Medical Advice and Information for people with Prader-Willi Syndrome (PWS)

*These 3 posters are left un-numbered to allow them to be displayed, but for the purposes of this booklet remain in sequential order.

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All website addresses were accurate at time of publication.

About this guide

This guide was written by medical specialists in PWS on behalf of the Prader-Willi Syndrome Association (PWSA) in the USA, a charitable organisation supporting individuals with PWS, their families and carers and professionals; and Consensus Gretton, an acknowledged specialist provider of support and accommodation for adults with PWS in the UK.

About Prader-Willi Syndrome

Prader-Willi Syndrome (PWS) is a complex neurobehavioural genetic disorder resulting from an abnormality on the 15th chromosome. It occurs in males and females and equally in all races.

PWS typically causes low muscle tone, short stature if not treated with growth hormone, cognitive deficits, incomplete sexual development, problem behaviours, and a chronic feeling of hunger that, coupled with a metabolism that utilises drastically fewer calories than normal, can lead to excessive eating and life-threatening obesity.

At birth the infant has a low birth weight for gestation, hypotonia, and difficulty sucking due to weak muscles ("Failure to thrive"). The second stage ("Thriving too well"), with onset between the ages of two and five throughout lifetime, may show increased appetite, weight control issues, and motor development delays along with behaviour problems.

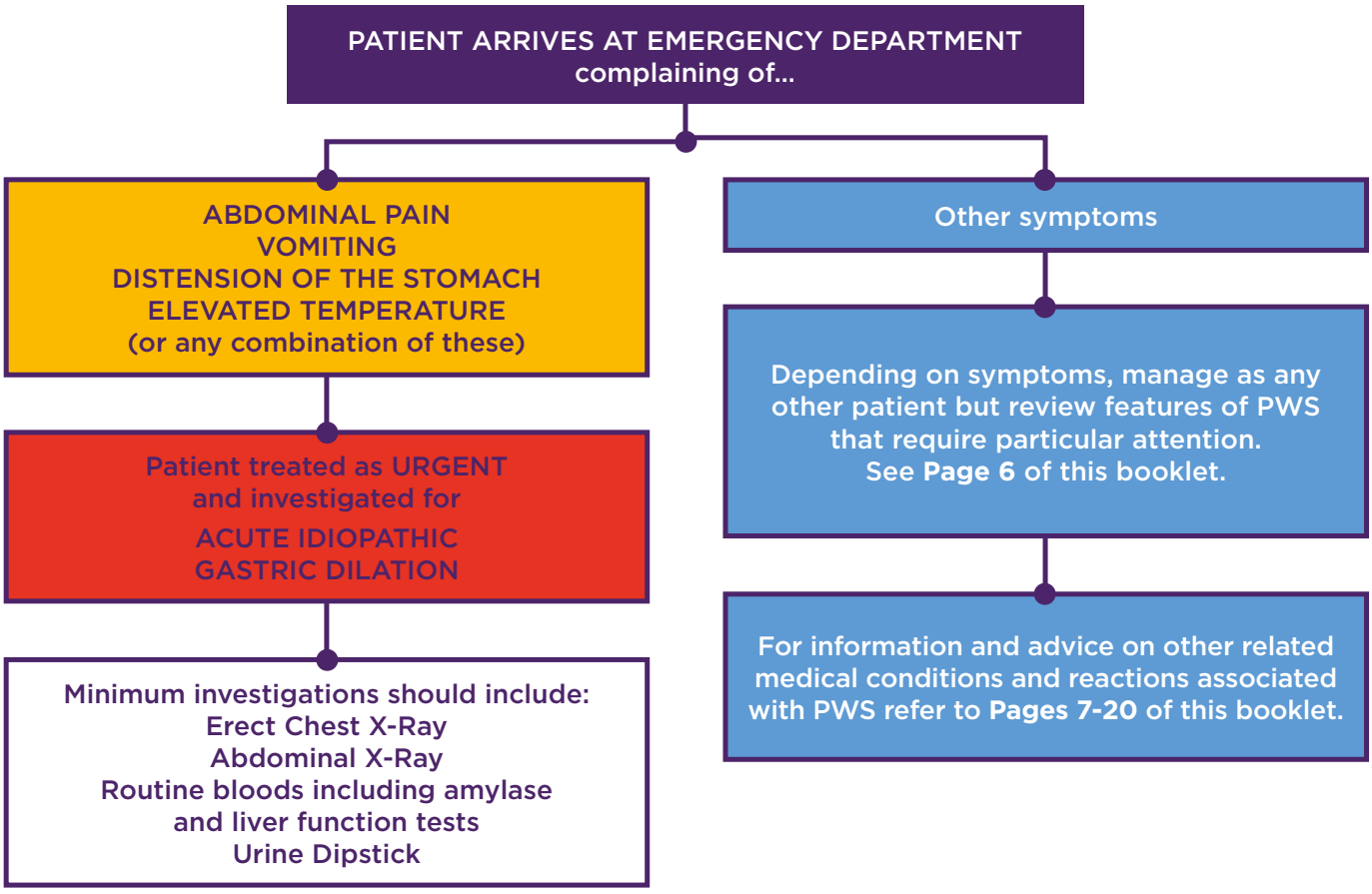
Other factors that may cause difficulties include negative reactions to medications, high pain tolerance, gastro-intestinal and respiratory issues, lack of vomiting, and unstable temperature.
Severe medical complications can develop rapidly in individuals with PWS.

Awareness of why symptoms are presenting is vital and could save someone's life

T: **0808 223 5320**
E: **enquiries@consensussupport.com**
W: **www.consensussupport.com/pwsservices**

PROCEDURE FOR THE TREATMENT OF INDIVIDUALS WITH PWS AT THE EMERGENCY DEPARTMENT

The following procedure is recommended for individuals with Prader-Willi Syndrome (PWS) on arrival at the Emergency Department:



MEDICAL ALERT – Anaesthesia, medication reactions

Individuals with PWS may have unusual reactions to standard dosages of medication/anaesthetic agents.

USE EXTREME CAUTION with:

Medication that causes sedation – Prolonged and exaggerated responses have been reported.

Medication with antidiuretic effects – Some can lead to water intoxication.

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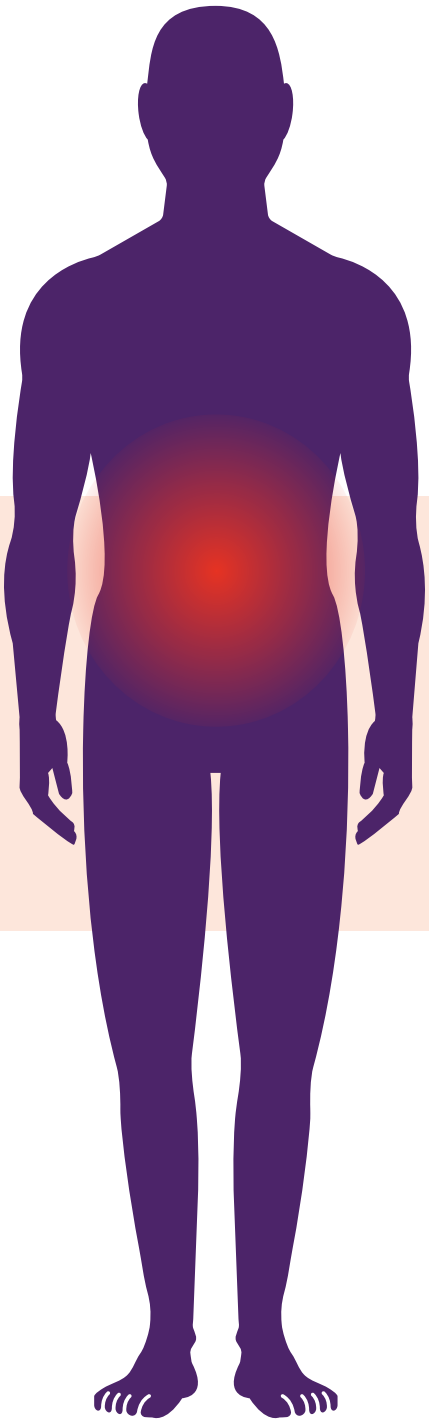
Information on people with
Prader-Willi Syndrome
for Medical and Healthcare Professionals

EMERGENCY PROCEDURE FOR PATIENTS WITH PRADER-WILLI SYNDROME (PWS)

NHS
Kettering General Hospital
NHS Foundation Trust

Acute Idiopathic Gastric Dilation is an important medical condition of which emergency services **MUST** be aware.

Although the condition is not common, it is more common in PWS than the general population. Acute idiopathic gastric dilation causes part of the stomach tissue to die in a similar way to how heart tissue dies in a heart attack. It has a very quick onset, is life threatening and may require emergency surgery to remove necrotic tissue.



A&E must respond urgently if people with PWS present with:

- **ABDOMINAL PAIN**
- **VOMITING**
- **DISTENSION OF THE STOMACH**
- **ELEVATED TEMPERATURE**

(Or any combination of these)

The patient should be treated as **urgent** and should receive the following minimum investigations as soon as is practically possible:-

- **Erect Chest X-Ray**
- **Abdominal X-Ray**
- **Routine bloods including amylase and liver function tests**
- **Urine Dipstick**

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Information on people with Prader-Willi Syndrome for Medical and Healthcare Professionals

Summary: PWS is a genetic disorder with documented patterns of health challenges and many hypothalamic related medical concerns

- Hyperphagia – excessive appetite from about the age of 2 but sometimes later
- Hypotonia – weak muscle tone especially in infancy
- Hypogonadism – most require hormone replacement
- Typically short stature and often growth hormone deficient
- Learning disability ranging from mild to severe
- Emotional problems including temper outbursts, anxiety 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

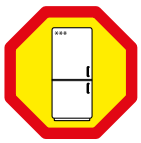
People with PWS do not normally vomit. When pain, flatulence, abdominal distension and vomiting are present, this could be life threatening. It may also be a sign that a large amount of food has been consumed, which can also be life threatening.

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 people 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 signal emergency GI issues.



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. Rectal prolapse may occur from a variety of mechanisms, lower extremity edema with venous stasis and vasculitis is associated with DVT and can cause fatal pulmonary embolism, 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 eg bruising, swelling or bone fractures. Do not expect the person with PWS to necessarily complain of pain whether ill or injured.

Anaesthesia

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 and 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 apnea is common.

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. Pulmonary embolism should be considered in anyone presenting with sudden cardiovascular compromise.

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.

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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.

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.

MEDICAL ALERT

Important Considerations for Routine or Emergency Treatment

Obesity and its related complications is the major cause of morbidity and mortality in Prader-Willi syndrome. Keeping the individual at a healthy weight will minimize these complications but there are important medical and behaviour problems unique to Prader-Willi syndrome regardless of weight status.

Central Adrenal Insufficiency in Individuals with Prader-Willi Syndrome

Several studies have shown CAI in individuals with PWS while others failed to show a deficiency. Stress dose of cortisol may be indicated if individual has problems after surgery or during times of stress. http://www.pwsausa.org and view Medical section under Adrenal Insufficiency.

Falls and Fractures

Individuals with PWS may have significant fractures from simple falls and require x-rays even if they do not complain of pain. Persistent pain, swelling, guarding, or decreased movement of the extremity for more than a few days may warrant an x-ray.

Hyperphagia (Excessive Appetite)

Individuals with PWS must be constantly supervised in all settings to prevent access to food. In hospital settings, obtaining unguarded food can lead to rapid ingestion and fatal choking. Individuals who have normal weight have achieved this because of strict external control of their diet and food intake; these individuals are not less likely to ingest available food. There are no treatments for this relentless hunger. Insatiable appetite may lead to life-threatening weight gain, which can be very rapid and occur even on a low-calorie diet.

Medications – Adverse reactions

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. Metabolism of the drugs may be impaired in individuals with PWS.

Pain Insensitivity

Lack of typical pain signals is common and may mask the presence of infection or injury. Someone with PWS may not complain of pain until infection is severe or may have difficulty localizing pain. Parent/ caregiver reports of subtle changes in condition or behaviour should be investigated for medical cause. Any complaint of pain by a person with PWS should be taken seriously.

Skin Lesions and Bruises

Because of a habit that is common in PWS, open sores caused by skin picking may be apparent. Individuals with PWS also tend to bruise easily. These lesions can cause serious life-threatening infections. There are approaches to help mitigate picking. http://www.pwsausa.org and view Medical section under Skin Picking. Appearance of such wounds and bruises may erroneously lead to suspicion of physical abuse.

Swallowing and Choking

Persons with PWS are highly likely to have an undetected swallowing problem that places them at risk for asphyxiation of a food bolus (choking), and they require a specific type of swallowing evaluation. A clinical or bedside evaluation is not sufficient to detect dysphagia in this population. They frequently cannot tell if they cleared their airway after swallowing, increasing the risk for aspiration. Choking can also occur with rapid ingestion of unguarded foods and has led to many deaths in the PWS population.

http://www.pwsausa.org and view Medical section under Choking/Swallowing.

Vomiting – Lack of ability to vomit

Vomiting rarely occurs in those with PWS. Emetics may be ineffective, and repeated doses may cause toxicity. This characteristic is of particular concern in light of hyperphagia and the possible ingestion of uncooked, spoiled, or otherwise unhealthful food items. The presence of vomiting may signal a life-threatening illness and may warrant immediate treatment.

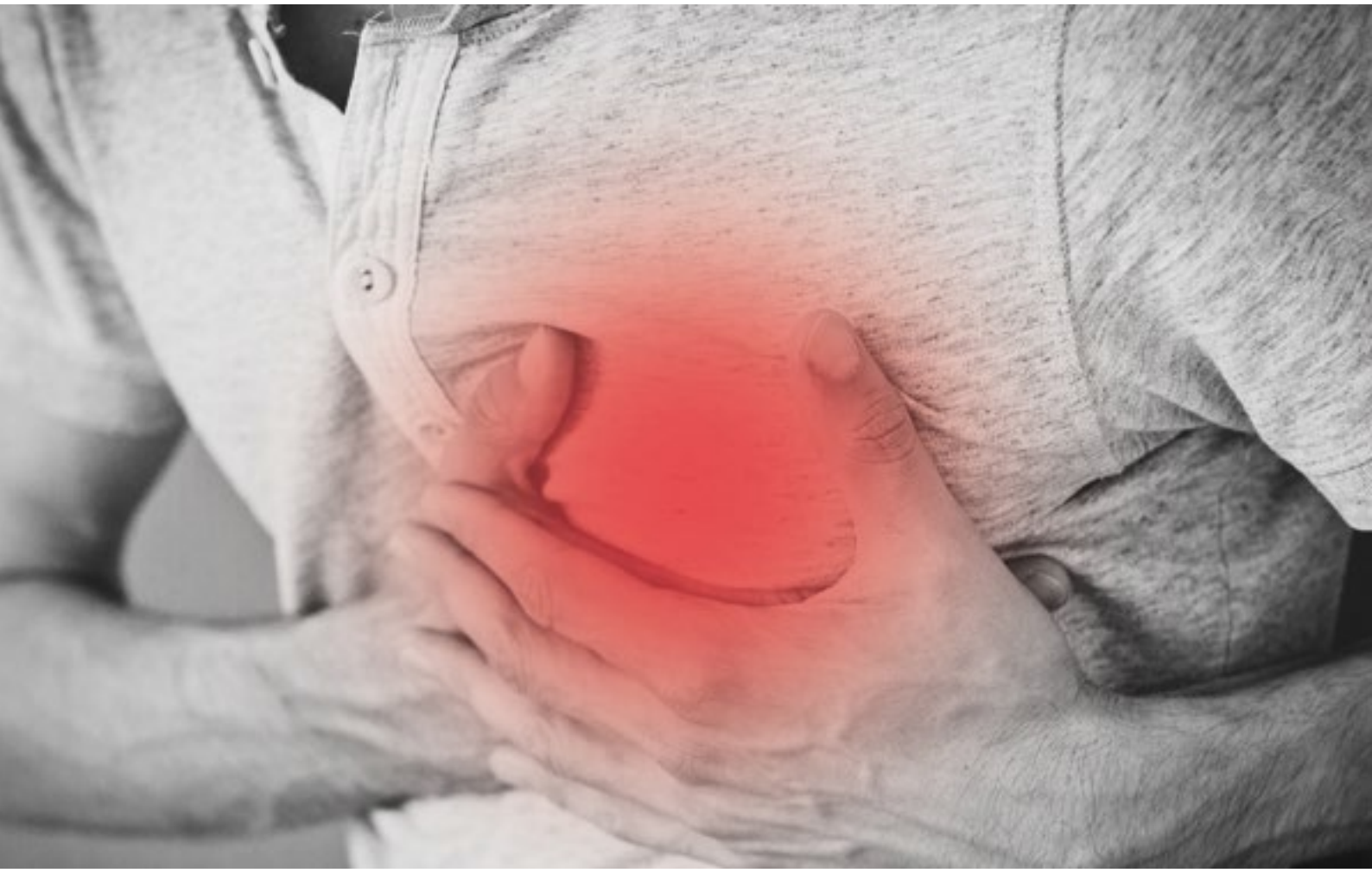
Water Intoxication

Water intoxication has occurred in relation to use of certain medications with antidiuretic effects, as well as from excess (binging) fluid intake alone. Anti-diarrheal medications may cause severe colonic distension, necrosis and rupture and should be avoided. http://www.pwsausa.org and view Medical section under Water Intoxication.

Respiratory Concerns

Individuals with PWS are at increased risk for respiratory difficulties. Hypotonia, weak chest muscles, swallowing abnormalities and sleep apnea are common. Anyone with significant snoring, regardless of age, should have a medical evaluation to look for obstructive sleep apnea. Infants commonly have central sleep apnea which generally improves over time but may also have obstructive sleep apnea due to hypotonia and other factors. Hypotonia can lead to diminished activity levels and low aerobic capacity.

Hypoventilation may be central in origin. In children with PWS, chronic stomach reflux and aspiration are emerging as common problems. Reflux should be considered in young children with chronic respiratory problems; videofluroscopy is the preferred test. Individuals with obstructive apnea or obesity are at more risk for reflux.



Recommendations for Evaluation of Breathing Abnormalities Associated with Sleep in Prader-Willi Syndrome

PWSA (USA) Clinical Advisory Board Consensus Statement – 12/2003

Problems with sleep and sleep disordered breathing have been long known to affect individuals with Prader-Willi syndrome (PWS). The problems have been frequently diagnosed as sleep apnea (obstructive [OSA], central or mixed) or hypoventilation with hypoxia. Disturbances in sleep architecture (delayed sleep onset, frequent arousals and increased time of wakefulness after sleep onset) are also frequently common. Although prior studies have shown that many patients with PWS have relatively mild abnormalities in ventilation during sleep, it has been known for some time that certain individuals may experience severe obstructive events that may be unpredictable.

Factors that seem to increase the risk of sleep disordered breathing include young age, severe hypotonia, narrow airway, morbid obesity and prior respiratory problems requiring intervention such as respiratory failure, reactive airway disease and hypoventilation with hypoxia. Due to a few recent fatalities reported in individuals with PWS who were on growth hormone therapy (GH), some physicians have also added this as an additional risk factor. One possibility (that is currently unproven) is that GH could increase the growth of lymphoid tissue in the airway thus worsening already existing hypoventilation or OSA. Nonetheless, it must be emphasized that there is currently no definitive data demonstrating that GH causes or worsens sleep disordered breathing. However, to address this new concern, as well as the historically well documented increased risk of sleep-related breathing abnormalities in PWS.

The Clinical Advisory Board of the PWSA (USA) makes the following recommendations:

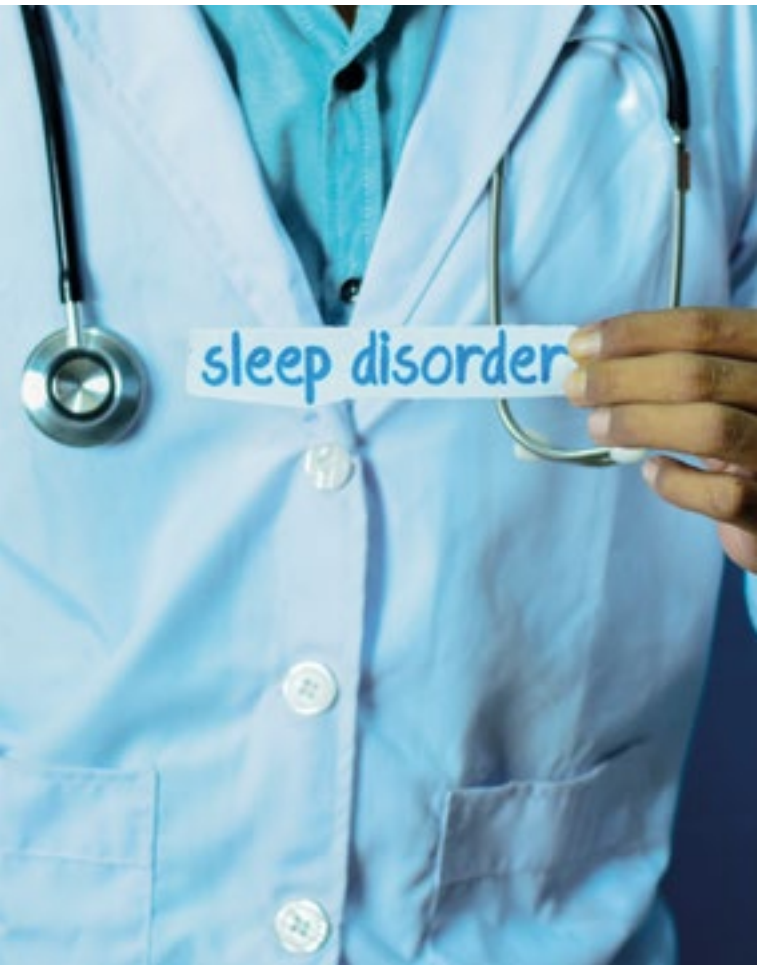
1. **A sleep study or a polysomnogram** that includes measurement of oxygen saturation and carbon dioxide for evaluation of hypoventilation, upper airway obstruction, obstructive sleep apnea and central apnea should be contemplated for all individuals with Prader-Willi syndrome. These studies should include sleep staging and be evaluated by experts with sufficient expertise for the age of the patient being studied.

2. **Risk factors that should be considered to expedite the scheduling of a sleep study should include:**

- Severe obesity - weight over 200% of ideal body weight (IBW).
- History of chronic respiratory infections or reactive airway disease (asthma). 11
- History of snoring, sleep apnea or frequent awakenings from sleep.
- History of excessive daytime sleepiness, especially if this is getting worse.
- Before major surgery including tonsillectomy and adenoidectomy.
- Prior to sedation for procedures, imaging scans and dental work.
- Prior to starting growth hormone or if currently receiving growth hormone therapy.

Additional sleep studies should be considered if patients have the onset of one of these risk factors, especially a sudden increase in weight or change in exercise tolerance. **If a patient is being treated with growth hormone, it is not necessary to stop the growth hormone before obtaining a sleep study unless there has been a new onset of significant respiratory problems.**

Any abnormalities in sleep studies should be discussed with the ordering physician and a pulmonary specialist knowledgeable about treating sleep disturbances to ensure that a detailed plan for treatment and management is made. Referral to a paediatric or adult pulmonologist with experience in treating sleep apnea is strongly encouraged for management of the respiratory care.



In addition to a calorically restricted diet to ensure weight loss or maintenance of an appropriate weight, a management plan may include modalities such as:

- Supplemental oxygen
- Continuous positive airway pressure (CPAP) or BiPAP
- Oxygen should be used with care as some individuals may have hypoxemia as their only ventilatory drive and oxygen therapy may actually worsen their breathing at night.
- Behaviour training is sometimes needed to gain acceptance of CPAP or BiPAP.
- Medications to treat behaviour may be required to ensure adherence to the treatment plan.

If sleep studies are abnormal in the morbidly obese child or adult (IBW > 200%) the primary problem of weight should be addressed with an intensive intervention - specifically, an increase in exercise and dietary restriction. Both are far preferable to surgical interventions of all kinds. Techniques for achieving this are available from clinics and centres that provide care for PWS and from the national parent support organization [PWSA (USA)].

Behavioural problems interfering with diet and exercise may need to be addressed simultaneously by persons experienced with PWS.

If airway related surgery is considered, the treating surgeon and anaesthesiologist should be knowledgeable about the unique pre- and postoperative problems found in individuals affected by Prader-Willi syndrome.

Tracheostomy surgery and management present unique problems for people with PWS and should be avoided in all but the most extreme cases. Tracheostomy is typically not warranted

in the compromised, morbidly obese individual because the fundamental defect is virtually always hypoventilation, not obstruction.
Self-endangerment and injury to the site are common in individuals with PWS who have tracheostomies placed.

At this time there is no direct evidence of a causative link between growth hormone and the respiratory problems seen in PWS. Growth hormone has been shown to have many beneficial effects in most individuals with PWS including improvement in the respiratory system. Decisions in the management of abnormal sleep studies should include a risk/benefit ratio of growth hormone therapy. It may be reassuring for the family and the treating physician to obtain a sleep study prior to the initiation of growth hormone therapy and after 6-8 weeks of therapy to assess the difference that growth hormone therapy may make. A follow-up study after one year of treatment with growth hormone may also be indicated.

Growth Hormone Treatment and Prader-Willi Syndrome

PWSA (USA) Clinical Advisory Board Consensus Statement - 6/2009

PWSA International Consensus Statement 2013

Both statements are found at **http://www.pwsausa. org** and view Medical section under Growth Hormone.

Inpatient Considerations

Access difficulties (venous and airway)

Obesity and poor muscle tone may complicate line placement. A small airway, high palate, and/or obesity may complicate ability to intubate. Saliva is often thick and sticky. Many persons with PWS will have difficult IV access due to increased fat mass and smaller than normal blood vessels. Outpatient procedures and general sedation may be especially problematic. Care must be taken during procedures done in out of hospital settings, and that proper equipment for resuscitation is immediately available and consideration for doing these procedures in an OR should be discussed. Procedures where more than light sedation is used may warrant an overnight observation.

Anaesthesia

People with PWS may have unusual reactions to standard dosages of anaesthetic agents. Use caution in giving anaesthesia. Serious problems occur during conscious sedation, if it is not well monitored, rather than from the use of general anaesthesia and airway management. Ongoing assessment of breathing and oxygen saturation is critical in all outpatient procedures including dental work. http://www.pwsausa.org and view the Medical section for articles on Anaesthesia.

- Anaesthesia and Prader-Willi Syndrome: James Loker, M.D., Laurence Rosenfield, M.D.
- Anaesthesia Concerns for Patients with PWS: Winthrop University.

Behaviour problems

Individuals are prone to emotional outbursts, obsessive-compulsive behaviours, and psychosis. Psychotropic medications may affect metabolism of anaesthesia leading to shorter or longer duration of action.

Cardiac problems

Surprisingly, coronary disease is less in PWS than in individuals with similar obesity. Cardiac problems usually are due to hypoventilation right heart failure. Edema can often be seen in the obese individual 16 even in the absence of heart failure and is treated by weight loss and ambulation. Diuretics are usually not that beneficial in treating the edema.

Cortisol Levels

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

Food seeking behaviours/ Relentless hunger

Complete safety from access to food is essential in any health care setting. Assume individual has eaten unless verified by caregiver. Complaints of hunger should not result in access to snacks or food. Patients in the hospital should have someone with them at all times. The individual may be on a caloric restricted diet and that should be conveyed to the nutritionist and kitchen.

Hypothalamic dysfunction – Pituitary deficiencies Hypothyroidism- Risk of central (TSH deficiency) hypothyroidism is 20-30% and may be undiagnosed prior to surgery.

Growth hormone deficiency - All individuals should be considered to be GH deficient.

Hypothalamic dysfunction is also the presumed origin of many other unique problems including temperature regulation, hunger, ventilatory effort, behaviour patterns.

Hypotonia

This muscle weakness may complicate ability to cough effectively and clear airways.

Narcotics

Individuals may have an exaggerated response to narcotics. Use the lowest possible dose to achieve the desired state of anaesthesia. Many individuals have delayed gastric emptying that can be compounded with narcotics.

Obesity

Consideration for obstructive apnea, pulmonary hypertension, diabetes, and right heart failure should be addressed.

Pain insensitivity

Unexplained tachypnea or tachycardia may be the only indication of pain. Behaviour problems which are not typical for this person may be evidence of pain. Individuals with PWS may not respond to pain in the same manner as others and it may mask the presence of underlying problems. Since pain may not be present, other signs of underlying problems should be monitored.

Pulmonary embolism

Individuals with PWS are at risk for pulmonary embolism. DVT prophylaxis should be considered in all obese individuals. Prolonged bed rest is to be avoided.

Psychosis

There is an increased risk of psychosis in individuals with PWS, which can be triggered by significant events such as changes in routines and serious illness. Prompt attention to hallucinations or reported change in typical behaviour is essential. View mental health issues http://www.pwsausa.org/medicalissues-a-z/ under Psychiatric concerns.

Saliva abnormalities

Thick sticky saliva complicates airway management especially during conscious sedation and increases the risk of caries. Dried saliva may not be an indication of hydration status. Water drinking is minimal in the majority of individuals with PWS.

Skin picking

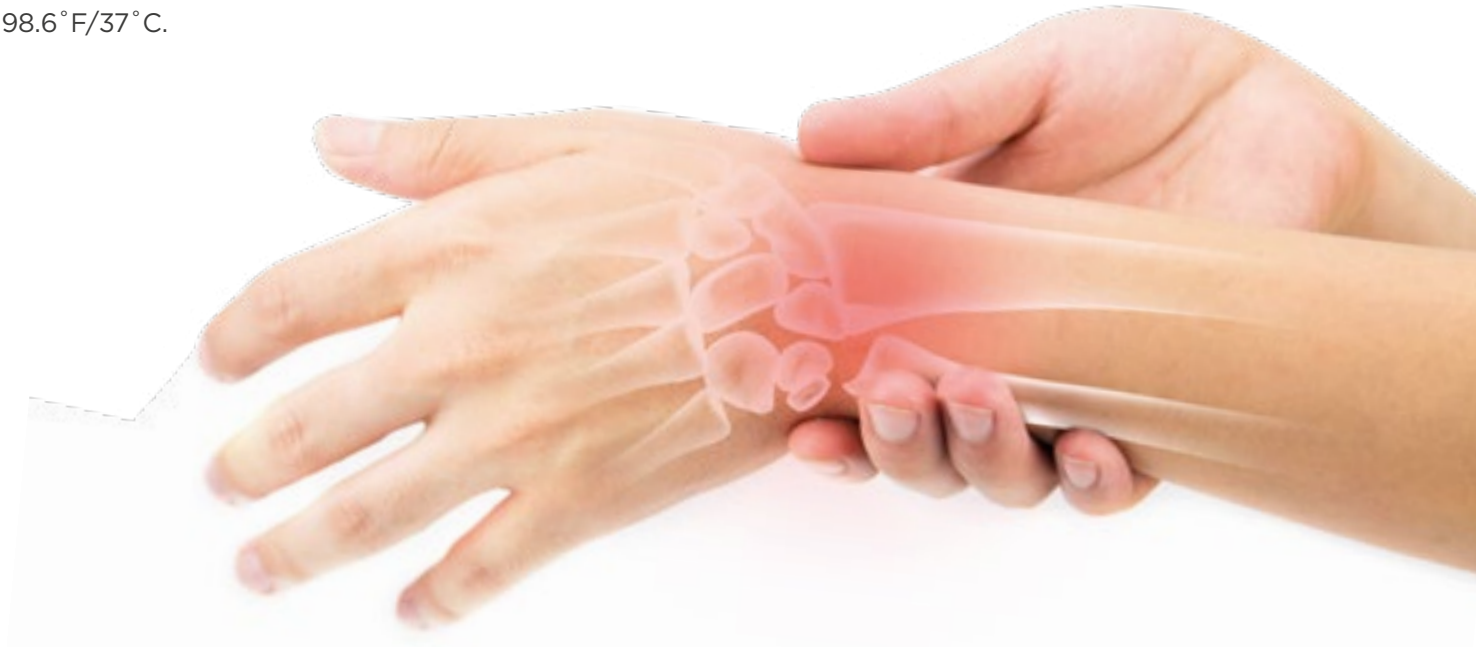
May complicate healing of IV sites and incisional wounds. Restraints or gloves may be necessary to protect wounds during healing.

Temperature abnormalities

Idiopathic hyper- and hypothermia have been reported. Hyperthermia may occur during minor illness and in procedures requiring anaesthesia. Fever may be absent despite serious infection. All individuals with PWS are at risk for mild hypothermia because of impaired peripheral somatosensory and central thermoregulation, poor judgment and cognitive inflexibility. Malignant hypothermia is a life-threatening problem occasionally seen in PWS. In cases of hypothermia, the patient may not report feeling cold. Baseline temperature may be below 98.6˚F/37˚C.

Surgical and Orthopedic Concerns

With the increasing number of infants and children with PWS undergoing sleep assessments prior to growth hormone treatment and the potential rise in surgical procedures (e.g., tonsillectomy) requiring intubation and anaesthesia, it will be important to alert the medical team about complications that may include trauma to the airway, oropharynx, or lungs due to possible anatomic and physiologic differences seen in PWS. They can include a narrow airway, underdevelopment of the larynx and trachea, hypotonia, edema, and scoliosis. Musculoskeletal manifestations, including scoliosis, hip dysplasia, fractured bones (which may be undetected), osteoporosis and lower limb alignment abnormalities, are described in the orthopedic literature. However, care of this patient population from the orthopedic surgeon's perspective is complicated by other clinical manifestations of PWS. http://www.pwsausa.org and view Medical section under Orthopedic Issues.



Post-operative Monitoring of Patients with Prader-Willi Syndrome

Patients with PWS are known to have increased morbidity after surgery due to:

- Abnormal physiological response to hypercapnia and hypoxia
- Hypotonia
- Narrow oropharyngeal space
- High incidence of central, obstructive and mixed apnea
- Thick secretions
- Obesity
- Increased incidence of scoliosis with decreased pulmonary function
- Prolonged exaggerated response to sedatives
- Increased risk for aspiration
- Decreased pain sensation
- Possible challenges with compliance to pre- and postoperative treatment procedures due to:
  - Extreme food seeking behaviour and hyperphagia due to hypothalamic dysfunction
  - High incidence of gastroparesis and slow motility of the intestinal tract
  - Extreme skin picking which may interfere with wound healing – Altered temperature regulation - fever may be absent in the presence of infection. There does not seem to be a higher incidence of malignant hyperthermia
  - The possibility of central adrenal insufficiency

Recommendations

- Patients with PWS who undergo deep sedation and general anaesthesia should be recovered overnight in a monitored unit. Infants and children may require intensive care monitoring.
- Continuous monitoring of pulse-oximetry for 24 hours postoperative with attention to airway and breathing.
- A conservative approach to pain management and use of narcotic agents.
- Full assessment of return of GI motility prior to initiation of intake by mouth because of the predisposition to ileus after surgery.
- Scheduling procedure as early in the day as possible to prevent prolonged time period where food seeking could take place.
- Direct supervision (1:1) to prevent foraging postoperatively.
- Monitor for picking at wounds and/or incisions. These may require additional dressings and other barriers including full time sitter to prevent access to surgical site and medical devices.
- Close observation of wound for signs of infection.
- Utilization of respiratory therapy interventions to prevent atelectasis and/or postoperative lung infection.
- Due to the hypotonia and obesity, individuals with PWS are at risk for deep venous thrombi (DVT) and pulmonary embolism. Patients should be under the guidelines for DVT prophylaxis. http://www.pwsausa.org and view Medical section under Postoperative Monitoring of Patients with Prader-Willi Syndrome.

Severe Gastric Intestinal Concerns

Vomiting (Lack of ability to vomit)

Vomiting rarely occurs in those with PWS. Emetics may be ineffective, and repeated doses may cause toxicity. This characteristic is of particular concern in light of hyperphagia and the possible ingestion of uncooked, spoiled, or otherwise unhealthful food items. **The presence of vomiting may signal a life-threatening illness and may warrant immediate treatment.**

Severe Gastric Illness

Gastric problems are very common in PWS due to decreased motility and gastroparesis. Abdominal distension or bloating, pain and/or vomiting may be signs of life-threatening gastric dilation, inflammation or necrosis. Rather than localized pain, there may be a general or vague feeling of being unwell. Any individual with PWS with these symptoms needs immediate medical attention. An x-ray, CT scan or ultrasound can help with the diagnosis and confirm if there is gastric necrosis and/or perforation.

If distension is noted, these individuals need close monitoring, made NPO and may need decompression with an NG tube.

Gastric necrosis or perforation is a medical emergency requiring exploratory laparotomy or emergent surgery. Individuals with PWS may not have tenderness, rigidity or rebound normally associated with an acute abdomen.

In addition to gastric distension, colonic impaction may also be present and need to be addressed. Stomach pain can also be due to gallstones or pancreatitis. An ultrasound, chemistry analysis of the blood and CT of the abdomen will help with the diagnosis.

Constipation in Individuals with Prader-Willi Syndrome

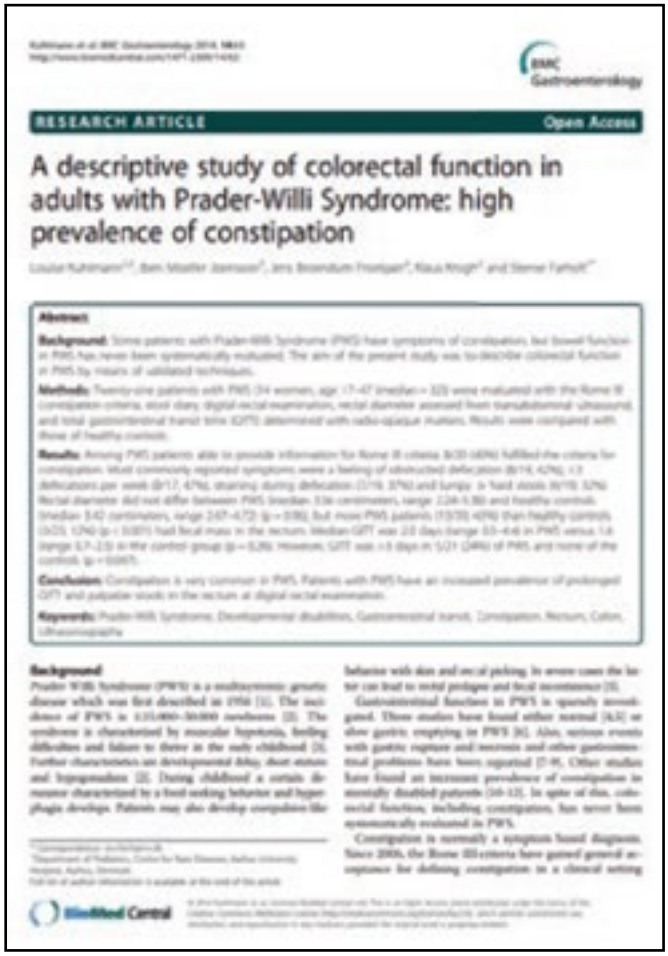
James Loker, M.D., Pediatric Cardiologist Ann Scheimann, M.D., M.B.A., Gastroenterologist PWSA (USA) Clinical Advisory Board Members

Constipation is a common problem in individuals with Prader-Willi syndrome (PWS). It takes longer for food to move through the GI system in Prader-Willi syndrome*. This slower passage of food can lead to serious issues similar to the ones seen related to the stomach. Outpatient methods used to clear constipation in non-PWS patients may be ineffective due to poor fluid intake and hypotonia. Inpatient regimens frequently use large volumes of fluid which may cause problems. Reliance on these methods may lead to life-threatening conditions such as necrosis and perforation of the colon and subsequent sepsis. Due to decreased muscle tone and altered pain response, individuals with PWS may not have the same clinical exam that a non-PWS patient would have. A heavier reliance on imaging may be necessary. Individuals with PWS may be at higher risk for impaction. Rectal examination and enema may be required in addition to oral cleanout regimen. This may also be problematic in some leading to rectal picking.

Patients with PWS having constipation and receiving repeated regimens of oral PEG (polyethylene glycol) solution for bowel cleansing should be monitored closely for abdominal distention and retention.

Failure of standard constipation protocols to clear the stool in a timely manner, especially in the face of increasing abdominal distension, vomiting, decreased appetite, stoppage of food consumption and/or abdominal pain warrants surgical or GI consultation. Emergent surgical or colonoscopic intervention may be necessary.

*Kuhlmann, et al. (2014) A descriptive study of colorectal function in adults with Prader-Willi syndrome: high prevalence of constipation. BMC Gastroenterology, Apr 4; Vol 14: page 63



RESEARCH ARTICLE Open Access

A descriptive study of colorectal function in adults with Prader-Willi Syndrome: high prevalence of constipation

Prader-Willi Syndrome (USA) ALERT! Risk of Stomach Necrosis and Rupture

Possibly Related to Chronic Gastroparesis

A Cause of Death from Sepsis, Gastric Necrosis or Blood Loss Signs and symptoms of stomach necrosis and rupture:

- **Vomiting** - Any vomiting is very unusual in Prader-Willi syndrome
- **Loss of appetite** - (ominous sign)
- **Lethargy**
- **Complaints of pain, usually non-specific** - Pain sensation is abnormal in Prader-Willi syndrome due to high pain threshold; rarely complain of pain
- **Pain** is often poorly localized
- **Peritoneal signs** may be absent
- **Abdominal/stomach bloating and gastric dilation**
- **Fever may or may not be present**
- **Temperature regulation** is altered in Prader-Willi syndrome
- **Guaiac positive stools (chronic gastritis)**

An algorithm for ER evaluation of an individual with PWS and abdominal complaints is on a foldout page in the back of this publication.

These Signs should raise suspicion of STOMACH NECROSIS/RUPTURE as a possible diagnosis which can be LIFE-THREATENING!

History may include:

- **History of binge eating within the week**
- **Hyperphagia and binge eating are characteristic** of Prader-Willi syndrome, regardless of whether obese or slim.
- **Frequently occurs** after holiday, or social occasion with less supervision of intake.
- **History of gastroparesis** - Common in Prader-Willi syndrome, though often undiagnosed.
- **Often slim or history of significant obesity followed by weight loss** - May leave the stomach wall thinned **http://www.pwsausa.org** and view Medical section under Gastric/Intestinal.

Consensus Gretton would like to thank PWSA (USA) for their help in producing this booklet. www.pwsausa.org

Consensus Gretton is part of Consensus who support over 650 adults and young people with learning disabilities, autism and complex needs.

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