**What is Prader-Willi Syndrome?**

**Major characteristics:** hypotonia, hypogonadism, hyperphagia, cognitive impairment, challenging behaviors

**Major medical concern:** morbid obesity

Prader-Willi syndrome is a disorder of chromosome 15 with prevalence: 1:12,000-15,000

**Obesity and its related complications** is the major cause of morbidity and mortality in Prader-Willi syndrome but there are GI problems unique to Prader-Willi syndrome that cause significant morbidity and mortality.

**GI complaints in PWS can be life threatening** and should be taken seriously. A recent study noted GI complications (obstruction, perforation, and distension) was the cause of death in 10% of the 312 individuals with PWS in PWSA (USA)’s study on deaths.

**Poor feeding and failure to thrive** in infants is generally short and G tubes are usually not indicated. Care should be taken not to limit calories in infancy. Dysphagia with thickened saliva, poor pharyngeal clearance and decreased esophageal motility place individuals with PWS at high risk for aspiration and choking deaths.

**Gastroparesis** (decreased GI motility), GI obstruction and constipation have been well documented in the syndrome. Lack of satiety in PWS can cause individuals to eat massive amounts of food which coupled with poor motility can cause massive gastric distension and perforation.

Individuals may present with decreased appetite, vomiting, abdominal distension/ pain, lethargy, or foul smelling belching with or without known eating binge. Some individuals have a high pain tolerance and with the

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**Prader-Willi Syndrome is:**

- A serious, life-long, and life-threatening medical condition
- One of the most common conditions seen in genetic clinics
- The most common genetic cause of obesity

**A major medical concern is morbid obesity.**

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**Prader-Willi Syndrome Association (USA)**
8588 Potter Park Drive, Suite 500
Sarasota, FL 34238

**Tel:** 800.926.4797 or 941.312.0400
**Fax:** 941.312.0142
**E-mail:** info@pwsausa.org
**Web:** www.pwsausa.org

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**Physician’s Resource for GI Emergencies in Prader-Willi Syndrome**

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**X-ray showing massive gastric distension in individual with PWS**
Evaluation of Individuals with Prader-Willi Syndrome with GI Complaints

Known or suspected binge, vomiting, lethargy, or abdominal pain/distension
Admit Patient for Observation

- NPO, and place NG. Urgent surgery consultation, CT abdomen
- Rectal exam
- Consider CBC, cortisol, CMP, amylase, lipase, lactate, ABG, PT/PTT, UA, BG

Is Patient stable and in no distress?

- NO
- Evidence of gastric necrosis or perforation?

- YES
- Constipation
  - Stool cleanout regimen
  - Check for impaction either digitally or with fleet enema prior to starting oral cleanout
  - Calorie restricted clear liquid diet
  - Check TFTs, electrolytes
  - Consider GI or surgical consultation, especially if stool guaiac positive
  - Reassess plain film in 12-24 hours
  - Increasing distension or failure to clear stool may indicate impaction or obstruction. Further GI consultation should be considered

- Gastric distension
  - NPO
  - NG for decompression
  - Make surgery aware of Patient
  - Reassess plain film in 6-8 hours unless condition worsens then urgent CT
  - Consider GI consultation, especially if guaiac positive stool

Is there evidence of gastric distension or constipation/impaction/obstruction?

- YES
- Short-acting anticholinergic medications
- Neostigmine 1 mg IM q6h
- Oral PEG 30-60 ml twice daily

- NO

Neither

- Check for UTI, pancreatitis, cholelithiasis, etc.
- RUQ ultrasound
- If patient vomiting consider esophageal obstruction or toxic ingestion

Recommended NPO and KUB flat plate and upright
- Rectal exam for stool guaiac

Emergent Surgery
- There is no advantage to waiting for surgery.

- NO
- Admit to ICU for observation
- Surgical consultation
- Check plain film every 6-8 hours unless examination changes
- Assess for other etiologies

NOTE:
Individuals with Prader-Willi syndrome have decreased muscle tone and pain response so clinical evaluation for acute abdomen is altered similar to a patient on steroids.

Patrick, age 21, died from a G.I. perforation.

Hypotonia may not present with a typical acute abdomen. The gastric distension can occur in thin individuals more frequently than obese and can happen in children as well. (see algorithm for management)

**Constipation** is a common complication in PWS. Outpatient methods to clear constipation may be ineffective due to poor fluid intake and hypotonia. Individuals may be at higher risk for impaction. Rectal examination and enema may be required in addition to oral cleanout regimen. Patients requiring repeated regiments of oral PEG solutions for bowel cleansing should be monitored closely for abdominal distension and retention.

Constipation may cause overflow loose stools and may be mistaken for diarrhea. Antidiarrheal medications can cause severe distension with perforation and death and care must be taken in patients with history of constipation and new onset diarrhea.

**Post-operative ileus** can be a significant problem that can delay recovery and be life threatening. Preoperative bowel program is advised. Individuals with PWS are more susceptible to narcotic effects and diet should be advanced carefully with frequent abdominal radiographs.

An algorithm (shown left) is included to help in management of the individual with PWS and GI complaints.

Medical professionals can contact PWSA (USA) to obtain more information and have a consult with a specialist, as needed.