Recognizing gastric dilatation and associated risks in Prader-Willi syndrome (PWS)

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A review of causes of death in PWS identified 8 cases resulting from gastric dilatation and necrosis (Stevenson et al, 2007). The clinical symptoms leading up to the terminal event were not available in this case series, but the data served to establish that people with PWS are at exceptional risk. This condition was first reported in a case series of 7 PWS patients by Wharton (1997). In this series, 2 patients with this condition died; 3 were treated surgically (subtotal or total gastrectomy); and 1 was treated conservatively with resolution of symptoms. In a case series identified by the authors, 8 patients with PWS aged 4-23 years suffered one or more episodes of gastric dilation, and in one person, the event resulted in death. In this case series we endeavored to obtain as much clinical detail related to the earliest signs and clinical course of the condition until definitive intervention was performed. Recent weight loss did not appear to be a predisposing factor in contrast to a European series of adults that suggested that a repeated history of weight loss and gain was associated with increased risk (Schrander-Stumpel et al, 2004). In our series, binge eating or gorging was not a precipitating factor, although a **recent change of diet** (eating out) or **exposure to carbonated beverages** was reported. Predisposing factors included **constipation** (due to impaction) or **diarrhea (due to viral gastroenteritis**). Constipating medications were sometimes but not always present. A helpful warning sign of impending crisis noted by parents was the presence of odiferous eructation ("**stinky burps**"). Patients with PWS rarely reported visceral pain, but a few of them did appear to be "**out of sorts**." **Food refusal**, **abdominal distention**, **abdominal discomfort** and **breathing difficulty** were important warning signs. **Vomiting** of foul smelling, undigested food from previous meals, or dark vomitus was an ominous sign, because vomiting is rarely seen in the PWS phenotype. These signs can occur without reports of nausea, pain or objective evidence of acute distress. Typical signs of an acute abdomen, such as rigidity, tenderness, and rebound, may not be present. Signs of inflammation or infection, such as fever or changes in vital signs may be absent. **Progression to necrosis and perforation appeared within 24-48 hours of presenting symptoms**, so prompt recognition and emergency management is essential.

Successful management includes an immediate **abdominal X-ray** that may indicate a huge distended stomach and proximal bowel with air fluid level. Fecal impaction may be an associated radiographic finding. **NPO** status and **decompression of stomach contents with a nasal gastric tube** is life-saving. IV fluid support is necessary. Emergency CT scan of the abdomen, GI consultation for gastroscopy, or surgical consultation may follow. Colonic cleanout from below may be indicated. Time until full resolution of the episode is highly variable, and careful monitoring throughout the duration of the episode is essential. After resolution of symptoms, refeeding must occur in the hospital with gradual advancement and close monitoring. ***A PWS person's communication of a desire to eat should not be viewed as an indication of symptom resolution.*** Recurrence of gastric dilatation has occurred under the circumstances of refeeding with inadequate follow up.

In the case of suspected gastric outlet obstruction, extreme caution must be exercised while performing aggressive gavage due to the risk of exacerbating gastric dilatation. Phenotypic hypotonia is an underlying risk factor predisposing people with PWS to this phenomenon. When gastroparesis occurs, dilatation to the point of necrosis (due to venous congestion) or rupture has occurred (Turan et al, 2003).

Medical management can be problematic due to the clinician's unfamiliarity with the medical and behavioral characteristics of PWS. Important features include the patient's difficulty in accurately experiencing and reporting abdominal discomfort due to extremely high pain tolerance and inability to localize the source of pain. Inflammatory response is similarly diminished in PWS. Behavioral issues, including minimization of discomfort and refusal of interventions, can cause misinterpretations of symptoms and can greatly delay or complicate management. Usually, successful recognition and timely action rely heavily on advocacy by parents, familiar caretakers, PCPs, as well as members of the PWSA Clinical Advisory Board.

Autopsy findings in the single death in this case series showed necrotic gastric tissue, gastric perforation and peritonitis. In the Stevenson and Schrandel-Stumpel reports, autopsy findings included tissue paper thin stomach wall, the etiology of which is uncertain.

Bibliography:

Schrander-Stumpel CT, Curfs LM, Sastrowijoto P, Cassidy SB, Schrander JJ, Fryns JP. Prader-Willi syndrome: causes of death in an international series of 27 cases Am J Med Genet A 2004 Feb 1;124A(4):333-8.

Stevenson DA, Heinemann J, Angulo M, Butler MG, Loker J, Rupe N, Kendell P, Cassidy SB, Scheimann A. Gastric rupture and necrosis in Prader-Willi syndrome. J Pediatr Gastroenterol Nutr. 2007 Aug;45(2):272-4.

RH. Wharton, Wang T , Graeme-Cook F, Briggs S , Cole RE. Acute idiopathic gastric dilatation with gastric necrosis in individuals with Prader-Willi syndrome Am J Med Genet 1997Volume 73, Issue 4, pages 437–441.

Turan M, Şen M, Canbay E, Karadayi K Yildiz E: 2003 Gastric Necrosis and Perforation Caused by Acute Gastric Dilatation: Report of a Case Surgery Today Volume 33, Number 4, 302-304.