

## ACUTE GASTROINTESTINAL EPISODES CAN BE LIFE-THREATENING

By Janalee Heinemann - Prader-Willi Syndrome Association (USA)

I recently received a call from a physician who told me that one of our mothers brought our Medical Alert articles with her to the emergency room. He said, "If she had not brought the articles and insisted I go to your web site, this child would have died. This information saved her life." His patient, a slim 15-year-old, had had an episode of binge eating. She came in with vomiting and belly pain. The physician said that normally he would have treated it like the flu for a couple of days. Due to our alerts, they pursued this further and found the girl with PWS had such a bad hernia that her spleen, stomach, and duodenum were in her chest. She is now recovering from surgery.

Unfortunately, not all parents carry the articles with them and not all physicians heed our warnings. In another recent situation, a slim young man had an episode of binge eating, and the ER and hospital did not take his symptoms seriously enough, soon enough. Even though we had one of our physicians called as a consultant and emphasized the urgent need for exploratory surgery, there was a fourteen to sixteen-hour delay in surgery before the local hospital physician believed how life threatening his condition was.

This young man had been doing very well prior to this incident and, a few hours after the eating episode, initially only exhibited signs of stomach pain and vomiting. See Dr. Rob Wharton's article which was initially printed in *The Gathered View* in 1999. What Dr. Wharton described was "acute idiopathic gastric dilation." This is where part of the stomach tissue dies, which is similar to a heart attack where part of the heart tissue dies. It comes on suddenly, is very life threatening, and needs immediate surgery. I have been speaking to several people, including our GI specialist, Dr. Ann Scheimann, and the pathologist who did this report with Dr. Wharton (who is now deceased) about the cause. Our conjecture is that if a person with PWS greatly distends their stomach with food (slimmer people may be more at risk) and does not get the normal message of full or pain, they may distend it to the point that it cuts off the blood supply thus causing necrosis. (The stomach becomes blackened and dead.)

Another risk of binge eating that can create a serious medical emergency is GI perforation. This may be the consequence of gastric necrosis. In addition, when there is severe stomach pain, a physician should consider an ultrasound due to the possibility of gallstones and pancreatitis. The pancreatitis can be differentiated by chemistry analysis of the blood and a CT of the abdomen.

### **Medical Alert: Stomach Problems Can Signal Serious Illness**

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We have recently recognized and reported\* an important medical condition in individuals with Prader-Willi syndrome which families and other care providers should know more about. Although the condition is not common in individuals with PWS, it is much more common in these individuals than in anyone else. It is important to recognize the condition because it can cause severe medical problems when diagnosis and treatment are delayed. The condition can be successfully managed, however, when recognized in a timely fashion.

We have called the condition acute idiopathic gastric dilatation. The condition often begins suddenly in individuals in their late teens or older. There is generally no known cause. The first symptoms of illness are vague central abdominal discomfort or pain and vomiting. Bloating of the abdomen, caused by swelling or distention of the stomach, may also appear at this time. The person's temperature may also begin to become elevated at this point. In addition, the individual often begins to look and feels quite ill.

Individuals in whom these symptoms appear should receive immediate medical attention:

- abdominal pain,
- bloating or distention, and/or
- vomiting

A simple X-ray or CT scan of the abdomen should be taken to look for abdominal distention. If abdominal distention is present and the individual has pain but is relatively well appearing, a test called an endoscopy should next be performed to test the person's stomach lining for signs of inflammation. If the individual has distention on X-ray and is quite ill, emergency surgery might be necessary to more closely examine the person's stomach for signs of inflammation and necrosis [death or decay] of the tissue lining the stomach wall. When severe distention and necrosis is present, treatment consists of surgical removal of a significant portion of the stomach.

*\* Wharton RH et al. (1997) Acute idiopathic gastric dilation with gastric necrosis in individuals with Prader-Willi syndrome. American Journal of Medical Genetics, Dec. 31; Vol. 73(4): page 437-441.*