Stomach and Intestinal Problems in Persons with PWS Some Answers . . . Many Questions

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We continue to learn more about health issues and Prader-Willi syndrome (PWS). Over the past few years, we have gained a greater understanding of various stomach and intestinal problems that appear to be somewhat more common in persons with PWS. We now know that symptoms of stomach distention or bloating can possibly be related to a condition called gastric dilatation, a life threatening condition. We may be beginning to have answers to other stomach and intestinal concerns; however, at the same time this knowledge is opening the door to more questions. It is our hope that we will gain a better understanding of these problems today so we can prevent them from ever occurring in children and adults with PWS tomorrow.

Gastroparesis, Gastric Dilatation/ Necrosis and PWS - What Do We Know?

PWSA (USA) receives calls from around the country about people with PWS who are experiencing acute gastrointestinal problems. More and more children are being diagnosed with a problem called GASTROPARESIS (weakness of the stomach). This condition occurs when there is a delay or slowing in the contraction of the stomach. Because of this delay, stomach contents build up and abdominal distention can occur. The stomach is a muscle that contracts very much like our heart muscle. Unlike the heart, instead of pushing blood, the stomach pushes food out of its cavity into our intestine for further digestion. Feeling full is our body's mechanism for regulating the amount of food that the stomach can accommodate. When a person overeats, the stomach stretches. It may become "over stretched" or distended. We know that persons with PWS do not have the normal mechanism of registering fullness as they eat. They are at a very high risk of over distending their stomachs.

It is believed that if persons with PWS greatly distend their stomach, it can stretch to the point that it cuts off its blood supply, causing necrosis (the stomach tissue dies). This can be a life threatening condition if it is not quickly diagnosed and treated. Over the past few years, it has been discovered that many people with PWS have developed ACUTE IDIOPATHIC GASTRIC DILATATION WITH GASTRIC NECROSIS. Unfortunately, most of these cases have been diagnosed in a postmortem examination.

Acute = Sudden onset/severe **Idiopathic = Exact cause unknown** **Gastric = Stomach**
Dilatation = Expand, stretch, open **Necrosis = Death of tissue (stomach)**

Persons with PWS may be at higher risk for having gastroparesis. Some of the risk factors seen in both conditions are summarized below.

Risk factors for Gastroparesis	Risk Factors Seen in Persons with PWS
Diabetes – most common cause	Diabetes – many persons with PWS have diabetes.
Adrenal and thyroid gland problems	Many persons with PWS have been found to have low functioning of their thyroid gland.
Certain drugs weaken the stomach – many antidepressants and heart medications	Many persons with PWS take antidepressant medications as part of behavior management and some may be taking heart medications.
Neurologic or brain disorders such as Parkinson's, stroke and brain injury	We continue to learn the effects of PWS on brain functioning.

It has been found that people with PWS who have suffered from acute idiopathic gastric dilatation with gastric necrosis have had this occur shortly after a binge episode. It is not surprising to learn that persons with PWS who already have generalized low muscle tone may have poor muscle tone in internal muscles of their body.

The usual symptoms seen in gastroparesis include abdominal distention or bloating, abdominal pain, heartburn, vomiting and regurgitation of stomach fluid into the mouth. These symptoms can be very difficult to detect in persons with PWS. Any signs of acute abdominal illness should be evaluated by a health care professional.

If a person with PWS is experiencing gastrointestinal symptoms and problems, he/she may be referred to a specialist called a gastroenterologist (a doctor who specializes in disorders of the stomach and intestine), who will conduct tests to determine the cause of these problems. Optimally, gastroparesis is diagnosed through a gastric or stomach emptying test. Food that has been "marked" is given to the patient. A scanner then tracks the time it takes for food to leave the stomach. Another test that may be performed is an electrogastrogram (EGG). This is a test similar to the EKG test done on the heart. The EGG measures the electrical

waves that normally sweep over the stomach with each contraction. In gastroparesis, these electrical waves are slower than normal.

If caught early, gastroparesis can be treated. If there is an underlying medical condition, this needs to be treated. Diet and nutrition must also be adjusted. Since fats delay stomach emptying, foods high in fats should be avoided. High fiber food also stays in the stomach for a long period of time. They may need to be restricted if gastroparesis is severe. Liquids leave the stomach faster so they are encouraged. It has also been found that eating frequent small feedings 4-6 times a day may be helpful. In many cases, medications may be used to help stimulate the stomach to contract and empty more normally. It is important to follow the advice and recommendations of the health care professional and/or dietician that are most knowledgeable of that person's condition and needs.

QUESTIONS TO BE RESEARCHED: Is gastroparesis a common finding in children and adults with PWS? What should be done to diagnose and treat this condition? Are there any persons with PWS who seem to be at higher risk?

Many parents and caregivers also report that children or adults with PWS experience rumination (the regurgitation of undigested food from the stomach back up to the mouth). For so long, it was believed to be a behavioral issue. Could this problem be related to gastroparesis? Do we need to change our approach in its management? Is this problem a sign of gastroparesis?

Constipation – Could this Be a Problem for Persons with PWS?

Over the past year, PWSA (USA) has been receiving an increased number of reports of constipation in children and adults with PWS. In many cases, this problem has been discovered by accident. Often, a large amount of stool has been noted in their intestines while having an x-ray or test done that is not specifically looking for this problem. So far, no research studies have been done to suggest or confirm if persons with PWS might be at higher risk. Intestines, like the stomach, are muscular organs which push its contents forward so that nutrients are broken down, absorbed and/or eliminated as part of the digestive process.

Constipation can be defined as infrequent passage of hard, dry stools or difficulty in evacuating stools. Ideally, a person should have a bowel movement every day or so, and it should be soft and bulky. There can be several causes for this problem. Some of the more common causes are summarized below.

Common Causes for Constipation
Lazy colon that does not contract properly and move the stool through the intestine (a "hypotonic" colon)
Thyroid deficiency
Low potassium level
Certain medications such as those used to manage mood/behavior, pain, diuretics (water pills)
Spastic colon
Tumors or advanced diverticulosis
Abuse of laxatives
Disruption in normal routine – often seen when a person travels

The longer stool remains in the colon, the greater the chance of it becoming hard and dry. As stool travels through the intestine, more and more water is absorbed, resulting in firmer, harder stool. When a person does not have adequate water intake, this can make this problem worse. Water and other fluids help to keep the stool moister and prevent this. If a person has a "lazy colon", it may be contracting; however, it may not be contracting strong enough to provide the person with adequate elimination of stool. In these situations, a person may be having a BM every day and still have a large quantity of stool remaining in their colon. This build-up can also cause over-distension of the colon. It can cause pressure – both forwards (toward the rectum) as well as backwards (toward the stomach). There has been a question as to whether this build-up of pressure in the colon may be adding to the problems in acute idiopathic gastric dilation and necrosis. Many persons who have suffered with this condition have also reported a problem with constipation.

The primary way constipation is diagnosed is by listening to a person's history and complaints. For many people with PWS, that reporting is often sketchy and incomplete. In the early years, parents may be assisting with toileting hygiene issues and see their child's stool. As they grow older and more independent, this is not the case. If a problem is suspected, the health care professional may perform a physical exam along with additional diagnostic testing. Blood testing may be done to rule out a thyroid or potassium deficiency. A barium x-ray may be performed; this is an x-ray using a contrast solution (barium) that is instilled with an enema into the lower intestine. Other tests such as a sigmoidoscopy or colonoscopy may be indicated. In both cases, a flexible lighted instrument is inserted into the rectum in order to view the intestine. The sigmoidoscopy allows the health care provider to view the rectum and lower descending colon. The colonoscopy is a more extensive test in which all of the large intestine can be viewed. If polyps (blood-filled growths that can often develop into cancer) are found, they can be removed and

biopsied. Growths may be one of the more serious causative factors that should be ruled out.

Treatment of constipation is dependent upon its cause. Once serious problems are eliminated, simple measures can be used.

Guidelines for Treating Constipation

- Eat regularly (not usually a problem for persons with PWS)
- Drink plenty of water and fluids daily (often difficult for persons with PWS)
- Encourage regular walking and/or exercise
- Use a bathroom when the urge to have a BM occurs. If needed, set aside 15 minutes after a meal to sit on the toilet.
- Eat a diet of high fiber, fruits and vegetables (fresh is often better)
- Use laxatives and/or enemas as recommended by your health care provider. There are different kinds of laxatives that work on the intestine in different ways. Your provider can assist you in choosing the correct one. Overuse of laxatives can cause the colon to become dependent upon them.

QUESTIONS TO BE RESEARCHED: Are persons with PWS at higher risk for constipation? Do they have a tendency to have a "lazy colon"? Are they effectively and adequately emptying the colon? Can people with PWS prevent issues with constipation? If yes, what appears to be the most effective form of treatment? Does constipation predispose the development of acute idiopathic gastric dilatation with necrosis?

Hemorrhoids and Anal Fissures – Could This Help to Explain Rectal Irritation and Picking?

Hemorrhoids are a common nagging disorder. They are dilated (enlarged) veins that occur in and around the anus and rectum. They may be internal (inside the rectal canal) or external (outside). They can cause some uncomfortable sensations and problems, including itching, irritation, bleeding and pain. If the hemorrhoids are external, they can often be seen as small protrusions from the anus. If they are internal, they may not be seen and a person may not be aware they have them. Conditions that can contribute to the development of hemorrhoids are poor bowel habits, constipation, diarrhea, obesity, pregnancy and straining during a bowel movement.

Anal fissures are small tears in the lining of the anus. They can result from a dry hard bowel movement that causes this tissue to break. They are also seen when a person experiences bouts of diarrhea or irritation. A fissure can be quite painful during and following a bowel movement. Bleeding and itching may also be associated with these. They can become infected so that an abscess or ulceration may develop. In such cases, fever, drainage and swelling may also be present.

Diagnosis of any rectal problem is primarily done by examination by a health care provider. Because of the presence of bleeding, further studies may be done to make sure other more serious conditions (colitis, Crohn's disease, polyps, and tumors) are ruled out. Many of these problems resolve with no treatment. However, symptomatic treatment is often helpful in minimizing the discomfort associated with these problems. Stool softeners help to reduce the pain in passing the stool. Medicated creams and/or pads may decrease pain and itching. A sitz bath (soaking the buttocks in warm water) helps to relieve discomfort and promote healing. Medications such as antibiotics and/or suppositories may be prescribed. The health care professional may need to surgically open up an abscess to promote drainage and relieve pain. The itching, pain and irritation of hemorrhoids and other rectal problems can be very bothersome. In addition to discomfort, the problem can be compounded if a person is also experiencing rectal pressure often associated with severe constipation.

QUESTIONS TO BE RESEARCHED: Do persons with PWS have a tendency to have problems with hemorrhoids, anal fissures or other rectal problems? Is the start of rectal picking a result of persons with PWS experiencing these sensations and/or problems? Could this problem be prevented or decreased by paying closer attention to bowel habits, hemorrhoids, anal fissures and/or other rectal conditions?

Questions and Answers - Where Do We Go from Here?

We are just beginning to question and learn how the gastrointestinal system works in persons with PWS. In addition, we are starting to gain knowledge about some of the health concerns that are also being diagnosed. We don't have all the answers. What we do know is that we need to take a closer look at stomach and intestinal problems in people with PWS. We must learn what can be done to prevent serious health problems from occurring. We must proceed by encouraging and supporting more research. We must do everything to nurture the search for more answers.