Pancreatitis in WAGR Syndrome
Information for Parents and Physicians

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People with WAGR syndrome are at increased risk for a condition called Pancreatitis. Pancreatitis is an inflammation of the pancreas, a large gland located behind the stomach. Pancreatitis is a serious condition. In most patients it causes significant pain, but this symptom may be absent in some individuals with WAGR syndrome. In rare cases, pancreatitis can be life-threatening.

What Is Pancreatitis?

The pancreas is an important organ. It releases insulin and glucagon into the bloodstream. These are hormones that help the body use the glucose (sugar) it takes from food for energy.

The pancreas also secretes digestive enzymes into the small intestines through a small tube called the pancreatic duct. These enzymes help digest fats, proteins, and carbohydrates in food. Digestive enzymes usually do not become active until they reach the small intestine, where they begin digesting food. If these enzymes become active while still inside the pancreas, they start "digesting" the pancreas itself, causing the organ to become inflamed.

There are two types of pancreatitis. Either can occur in patients with WAGR Syndrome:

- **Acute** pancreatitis occurs suddenly and resolves after a short period of time
- **Chronic** pancreatitis does not resolve, and can result in a slow destruction of the pancreas

Either type of pancreatitis can cause serious complications including bleeding, tissue damage and infection. In extreme cases there can also be damage to other organs.

What causes Pancreatitis?

There are several reasons why people with WAGR syndrome may be susceptible to pancreatitis:

- One copy of the PAX6 gene is deleted in most people with WAGR syndrome. The PAX6 gene is involved in the development of the pancreas. Deletions of this gene may result in defects in the anatomy or function of the pancreas
- Some people with WAGR syndrome have a condition called “hypertriglyceridemia.” Hypertriglyceridemia occurs when there are high levels of

fatty molecules in the blood, called triglycerides. It is thought that having high levels of triglycerides in the blood increases the risk of pancreatitis.

- Certain drugs can also cause pancreatitis. The medication that is associated with pancreatitis in WAGR syndrome is an anesthetic agent called Propofol. Propofol is frequently used for medical procedures requiring general anesthesia. ***Propofol should be used with caution in patients with WAGR syndrome who have elevated triglycerides***

Diagnosis

Symptoms of **acute** pancreatitis may include:

- Nausea
- Vomiting with or without diarrhea
- Upper stomach pain that may or may not penetrate to the back
- Fever
- Rapid pulse
- Anxiety
- Restlessness or irritability
- Tender abdomen upon examination
- Discoloration of bowel movements (yellow and/or gray)

During an **acute** attack, the blood contains high levels of lipase. Lipase is a digestive enzyme formed in the pancreas. A doctor can order a blood test to look for higher than normal levels of lipase, which may indicate that pancreatitis is present. The doctor may also order imaging tests, such as x-rays or CT or MRI scans.

In **chronic** pancreatitis, typical patients have abdominal pain. The pain may get worse when eating or drinking. The pain may spread to the back or become constant and disabling. Other symptoms may include nausea, vomiting, weight loss, oily stools, frequent bowel movements with passage of undigested food.

50% of patients with WAGR syndrome have an abnormally high tolerance for pain (the result of deletion of one copy of the BDNF gene). These patients may not exhibit signs of pain or discomfort, even when acute pancreatitis is present.

Individuals with WAGR syndrome who are nonverbal may have difficulty communicating their pain. These individuals may express pain or illness as a change in behavior. For example, they may become withdrawn, clingy, or aggressive.

Treatment

Treatment for **acute** pancreatitis usually involves medication for pain relief, placing the patient on nothing by mouth/intravenous fluids only, or on an all liquid diet, and gradually moving to a diet high in carbohydrates and low in fat. Antibiotics are sometimes also given.

Treatment for **chronic** pancreatitis may involve some or all of the same measures, along with pancreatic enzymes to be taken with meals if it is determined that the pancreas is not secreting enough enzymes on its own.

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