

Reflux — Managing a Common Concern

by Carol Potter, MD

As Part I of a two-part review of GI issues in persons with CdLS, Dr. Carol Potter shares her expertise in working with patients who have CdLS and gastroesophageal reflux disease (GERD).

Gastroesophageal reflux disease (GERD) is a common problem in persons with CdLS. By understanding more about what happens physiologically during reflux, what complications can occur, how it is diagnosed, and what treatments are helpful, parents and caregivers are better equipped to provide help for their children with CdLS.

Gastroesophageal Reflux Disease (GERD)

After food is eaten by mouth, it travels to the stomach through the esophagus (or “food tube”). Food then stays in the stomach to be mixed with acid produced by the stomach before moving into the intestine. Gastroesophageal reflux occurs when food that is in the stomach goes back up into the esophagus. All of us have some gastroesophageal reflux. For most people the episodes are short, and may not even be noticed. GERD becomes problematic when the episodes are long, exposing the esophagus to irritating acid, or when aspiration (stomach contents entering the lungs) occurs.

Complications of GERD

The esophagus has a delicate lining that is not made to withstand stomach acid. Severe reflux may have episodes that last from several minutes to more than an hour. This can lead to pain as the acid irritates the delicate lining of the esophagus. The source of this pain may be hard to identify even in children without developmental delay. It may be impossible for children with communication difficulties to tell us where it hurts. They may show their pain by acting out, not eating, or self-injurious behavior. However, some children may eat more as the food temporarily neutralizes stomach acid, making them feel better for a few minutes. The symptoms of GERD are not necessarily specific and may be similar to those caused by an ear infection or other illness. Thus, diagnosing GERD requires that doctors do some detective work.

Prolonged exposure of the esophagus to acid can also cause damage to the esophagus, leading to inflammation called esophagitis. The esophagus pushes food to the stomach much like we push toothpaste out of a tube. When the esophagus is inflamed from acid, the muscles are not coordinated and don't push the food down. Food may feel like it is stuck, and may indeed become stuck, in a wide-open esophagus because of this poor muscle coordination. This sensation is very frightening and can lead to poor eating. Long-term inflammation may also lead to stricture (narrowing of the esophagus by the presence of scar tissue which makes it too small for food to get through). Inflammation that goes on for years can lead to Barrett's esophagitis, an advanced stage of reflux where chronic peptic ulceration of the lower esophagus may occur. This injury to the esophagus can eventually lead to cancer.

Children with GERD may have poor growth rates. This can occur if they vomit much of their food. It can also occur because it hurts the children to eat, and they may cut back on what they would normally eat. If this pattern continues, the poor eating may continue once the pain is gone because it has become a learned behavior.

Aspiration (breathing things like food into the lungs) is a significant concern in children with CdLS and GERD. Children may choke and gag on food that comes back up to the mouth and some may get in their lungs. Wheezing, pneumonia, or chronic lung disease can result from aspiration. This complication of reflux is difficult to diagnose, treat, and prevent.

Diagnosis

I am often faced with the practical question, “Should I assume a child has GERD and treat it before I do test or should I test first?” Many children with CdLS have GERD, and it is often severe. Even if I test them for GERD, I usually have to repeat the test to make sure that the treatment I prescribed is working. I often assume a child has GERD and administer the test only after a few weeks of treatment. This is warranted due to frequent presence of GERD in children with CdLS and the special concerns of doing invasive tests on these children.

Many tests are used to diagnose GERD. The esophago-gastroduodenoscopy or EGD is probably the most useful. For this test the child is sedated (or given anesthesia) and a thin fiber-optic scope is passed through the mouth and into the esophagus, stomach, and duodenum. This allows us to see the tissue but more importantly, to biopsy each of the organs. The pinhead-sized biopsies allow us to see if there is inflammation, other tissue damage, or infection in the upper gastrointestinal tract. For the many children with CdLS whose GERD is severe and silent (they do not exhibit discomfort), this approach allows us to evaluate the extent of the process. This is a safe test to perform on children with CdLS, but not one I like to perform more times than I have to. Since there is often severe GERD injury on the pretreatment biopsy, I usually treat with therapy for 6 weeks and then do a follow-up biopsy to make sure that the therapy is adequate.

Upper GIs are sometimes used to look for reflux. In this test the child swallows barium and the radiologist watches the barium on x-ray. This will tell us if all the twists and turns are in the right place but this test won't tell us if there is tissue damage to the esophagus. Sometimes GERD can be seen, but an Upper GI is a sort of snap shot of what is going on in the stomach and esophagus and might not catch reflux activity that may be manifested at other times. The child is often scared and crying, and doctors have to twist the child around to see all the views; it is not a very accurate picture of the severity of the GERD. A modification of this test, the swallow study, looks at the mouth and upper esophagus only. This test can be very useful to any speech pathologist or occupational therapist who is helping the child with swallowing.

Sometimes pH probes are used in the diagnosis of GERD. This thin, rubber-coated wire with an acid meter on the end measures acid in the esophagus. This allows you to find out if the child has GERD and if it occurs at the time when the child has symptoms. It, however, cannot tell you if there is tissue injury. The probe has to stay in place for 24 hours and this can be problematic for a child with CdLS.

Treatments

Once the diagnosis of GERD is made, therapy should be tailored for the problems that the child is having. Medicine that prevents the child from making acid in the stomach is often given to relieve pain and heal inflammation. Examples of medications that help suppress acid include Zantac, Pepcid, Tagamet, Prilosec, Prevacid, and Nexium. Drugs that help the esophagus and stomach empty (motility) can also be given although they aren't always as effective. Medications that help motility include Reglan, Erythromycin, and Cisapride (available in the U.S. only at centers participating in a compassionate use program). Tube feedings are sometimes used to improve nutrition. If medical therapy fails, surgery (fundoplication) can be done in an attempt to keep food in the stomach. This is an extensive operation with a number of possible complications, so it is not used as first line therapy.

Due to the rate at which GERD occurs and despite its often silent nature in patients with CdLS, GERD should be evaluated in all patients with CdLS and aggressively treated when found. A Gastroenterologist is an important member of their care team.

GERD is only one of the GI conditions that people with CdLS may experience. Updated information about bowel obstruction, malrotation, and pancreatitis will be featured in Part II of our review of GI issues.

GI Distress — Diverse Manifestations

by Lynn P. Audette, M.S.W., and Bruce Cleinman, M.A., Family Service Coordinators

GI problems are among the most common and troublesome issues we discuss with parents on a daily basis within the Foundation. Surprisingly, the cause of much GI distress turns out to be reflux related. In our experiences with families, it has become increasingly evident that among the most vital evaluations that a child can have are those to detect whether or not reflux is an existing condition. Reflux is a hallmark of CdLS. Up to 85% of individuals with CdLS experience some period of reflux in their lifetimes (most commonly in infancy or beginning of adolescence). Gastro-intestinal (GI) issues have diverse manifestations as the Abbotts and Smisloffs illustrate.

Jan and Jim Abbott wrote to the Foundation describing their experiences with their son, Gregg, who is 38 years old and moderately affected with the syndrome.

“Initially, he had difficulty eating but the reflux symptoms disappeared at about 2 years of age. He enjoyed a healthy childhood, had an excellent appetite, and a happy disposition. His only surgery was a bilateral inguinal hernia operation at the age of 13.

Two years ago, acting on the Foundation’s recommendation, we requested that Gregg be checked for silent reflux as it is a common disorder for people with CdLS. He had no behavior problems and did not appear to have reflux symptoms but he was having a lot of dental problems. An endoscopy indicated significant esophageal damage and a bacterial stomach infection. Gregg now takes Nexium one hour prior to his evening meal each day and has a yearly endoscopy. His doctor takes throat biopsies and monitors the condition. Periodically, Gregg has required antibiotics for the stomach infection. We urge all parents of children with CdLS to have their children examined for reflux even if they appear healthy and have no apparent digestive symptoms. Gregg’s dental problems and esophageal damage might have been avoided with earlier detection.”

Beth Smisloff, mom to Will (age 12) felt it important to share her family’s experience in hopes of raising awareness about pancreatitis, a much less common GI condition that has affected other people with CdLS.

“Will became ill in April exhibiting signs of lethargy, pain, and self-injurious behavior which led us to believe he may have a bowel obstruction. X-rays and a CT scan performed in the Emergency Room (ER) were negative and Will was sent home with a diagnosis of constipation. When his condition did not improve overnight, we contacted his doctor again. He called the ER and asked them to reread the results. We were told to report back to the ER immediately, as they had found something previously undetected — possible bowel obstruction. Will was then admitted to the Pediatric Intensive Care Unit where 24 hours later blood tests, which checked the pancreatic enzymes, revealed that he had pancreatitis. During his one-week stay, Will was treated for pain management and was off food intake entirely, giving his pancreas a chance to rest. When he was discharged from the hospital, he had improved but was still not back to 100%.

We were told pancreatitis typically resolves within a week but Will’s symptoms and discomfort continued for the next few weeks. Repeat CT scans were found to be abnormal. We pressed to have him admitted to Boston Children’s Hospital where there were pancreatic specialists. A two-week stay and thorough testing revealed that he indeed had smoldering pancreatitis.

He was discharged and under treatment for his pancreas (a j-g tube was placed for 24 hour continual feeds which allowed his pancreas to further rest) and yet he continued to act as if he did not feel well. Behaviorally, he was out of control with massive head banging and hand biting. He was miserable most of the day, nearly every day. We were feeling at the end of our rope wondering if there was some other physical explanation at the root of the problem or if he was starting with adolescent behavior problems. We were just steps away from trying medication to address behavior or looking into residential placement.

In August, an endoscopy performed during a final trip to Boston Children's revealed an extensive yeast infection in his esophagus caused by a stricture at his fundoplication site. Tests also revealed that his pancreas was back to normal. Finally we had answers to the cause of his behavior problems. Four weeks after treatment of the yeast infection [with Diflucan after initially being unsuccessful with oral Nystatin] we finally had our old Will back. He was singing, twirling, laughing, and smiling again. There was no head banging or hand biting.

The CdLS Foundation message I always think of is 'make sure there are no medical explanations for behavioral differences.' In this case we persisted and didn't take 'no' for an answer until we were certain we had gotten to the root of the problem. It takes patience, persistence, and sometimes the boldness to be a strong voice in the ears of the medical community, but these are imperative characteristics we must adopt in order to provide for the health and well being of our children. It was a long and difficult five months for Will and our family, and it highlighted our feelings that some physicians don't always look to the gray areas because they are so used to going by the book. Our children don't always get sick 'by the book' and that needs to be stressed."

If undetected, GI issues can affect many other systems (behavior, dental, ear, and sinus). We are thankful to the Abbotts for once again reinforcing the message that all individuals with CdLS need to be evaluated for reflux and are grateful to the Smisloffs for broadening our perspective on additional GI conditions. We encourage you to contact us with your questions as we can provide extensive materials, information, and facilitate consultation with the GI specialists on our Clinical Advisory Board.