

QUICK FACTS ON GI CONCERNS:

What Families and Providers Should Know

CdLS Foundation

Children and adults with CdLS often face a range of medical challenges, and gastrointestinal (GI) problems are among the most common. Understanding the signs, diagnosis, and treatment options can help parents, caregivers, and healthcare providers recognize symptoms early and provide the best care possible. Below are some of the GI conditions most frequently associated with CdLS.

GI problems are not just uncomfortable; they can lead to serious health complications if left untreated. In CdLS, where communication challenges may make it difficult for individuals to describe their pain or discomfort, caregivers and clinicians must closely monitor physical signs, such as changes in feeding, weight, or breathing.

While GI complications can feel overwhelming, knowledge is empowering. By understanding the risks, recognizing the signs, and advocating for timely medical care, families and providers can help children and adults with CdLS live healthier, more comfortable lives.



Key Takeaways

- Trust your instincts. Persistent vomiting, weight loss, breathing issues, or unexplained irritability should always be taken seriously.
- Early diagnosis and intervention significantly improve outcomes.
- A team approach, including pediatricians, gastroenterologists, surgeons, and CdLS specialists, ensures comprehensive care.

GI CONDITIONS	COMMON SYMPTOMS	DIAGNOSIS AND TREATMENT
<p>Gastroesophageal Reflux Disease (GERD) is when stomach contents flow back into the esophagus. In CdLS, reflux can be persistent and severe, sometimes causing long-term damage to the esophagus, throat, or even the lungs.</p>	<ul style="list-style-type: none"> • Chest pain (non-burning, radiating to the back) • Difficulty swallowing (dysphagia) • Sore throat and chronic cough • Increased salivation • Shortness of breath 	<p>Doctors may use tests such as an upper endoscopy, pH monitoring, manometry, or barium swallow to confirm GERD. Treatments typically begin with lifestyle and dietary modifications, as well as the use of medications. In some cases, endoscopic procedures or surgery may be necessary, especially if other therapies don't bring relief.</p>
<p>Pyloric Stenosis is a condition that occurs when the muscular valve between the stomach and small intestine becomes abnormally thick, narrowing the passage and preventing food from moving forward.</p>	<ul style="list-style-type: none"> • Forceful vomiting after feeding • Constant hunger • Dehydration • Weight loss • Visible stomach contractions 	<p>An ultrasound is the standard diagnostic test. Pyloric stenosis requires surgery (pyloromyotomy) to loosen the muscle and allow food to pass normally. Before surgery, fluids and electrolytes are carefully replaced to prevent complications.</p>
<p>Diaphragmatic Hernia is an abnormal opening in the diaphragm that allows abdominal organs to move into the chest, crowding the lungs. This condition may be identified at birth or even before, through a prenatal ultrasound.</p>	<ul style="list-style-type: none"> • Severe breathing problems soon after birth • Bluish skin (low oxygen) • Rapid breathing and heart rate 	<p>X-rays and physical exams can confirm the condition. Surgical repair is required to reposition the organs and close the diaphragm opening. Infants usually need breathing support during recovery.</p>
<p>Intestinal Malrotation is a birth defect where the intestines don't rotate properly during fetal development. Volvulus is a twisting of the intestine, often associated with malrotation, which blocks the intestine and can cut off its blood supply.</p>	<ul style="list-style-type: none"> • Vomiting bile (green fluid) • Abdominal pain and swelling • Constipation or diarrhea • Bloody stools • Poor growth (failure to thrive) • Rapid breathing or heart rate 	<p>Tests may include abdominal X-rays, CT scans, barium swallow or enema, and sometimes flexible sigmoidoscopy. Treatment often requires IV fluids, antibiotics, and the insertion of a nasogastric tube. Surgery to untwist the intestine is urgent and often lifesaving.</p>