

GASTROINTESTINAL (GI) ISSUES in Individuals with CdLS

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Many individuals with CdLS experience gastrointestinal (GI) issues at some point, and parents often find themselves navigating feeding challenges from an early age. Some of the most common concerns include feeding difficulties, acid reflux, and constipation. The reassuring news is that these issues are treatable, and numerous tools are available to support a child's comfort and growth.

Sorting out what is typical for CdLS versus what may signal something more serious can feel confusing. Everyday symptoms, such as reflux or slow digestion, are common in the CdLS community. However, when an anatomical difference is present, such as intestinal malrotation or a diaphragmatic hernia, symptoms often look more intense. Persistent vomiting, signs of breathing trouble, or vomit that appears bright green (due to bile) can be important clues that further testing is necessary.

Some conditions, like volvulus, develop suddenly and require emergency care. While there's no way to predict a volvulus, parents can look out for severe belly pain, a swollen or hard abdomen, and ongoing vomiting that may also be bilious. Other complications, like aspiration pneumonia, can occur if food or stomach contents accidentally enter the lungs. Persistent cough, low oxygen levels, fever, or shortness of breath after vomiting are reasons to seek medical attention.

The connection between CdLS genetics and gut motility is still being studied; however, several medications can help keep digestion moving more smoothly.

The best approach depends on a child's specific symptoms, which is something families can explore with their GI team.



Looking long-term, the outlook for GI health is often positive. As children grow into adulthood, reflux is the symptom that most commonly continues. Long-standing reflux can irritate the esophagus; therefore, adults with CdLS may occasionally require an endoscopy to assess for inflammation or early changes that necessitate treatment.

Feeding difficulties are highly individual; there's no single cause in CdLS. Some children require additional time to develop the coordination necessary for chewing and swallowing. For others, discomfort from reflux or inflammation makes meals stressful. One condition seen more frequently is eosinophilic esophagitis (EoE), where food-related inflammation affects the esophagus without causing typical allergic reactions. Identifying the root cause allows care teams to tailor therapies in a way that feels achievable for families.

Therapy and behavioral support can also help when food aversions or mealtime anxiety become barriers to nutrition. For some families, working with a feeding psychologist, alongside therapists, provides the right mix of strategies and understanding.

Supportive therapies can also make a meaningful difference. **Speech therapy** is helpful for children who are working on oral-motor coordination or have both feeding and speech delays. **Occupational therapy** can help when sensory challenges, like food textures or fine motor skills, make eating difficult. Many children benefit from a combination of both.

Sometimes, despite therapy and medication, growth still doesn't progress as expected. In those moments, a **feeding tube may become part of the plan.** Tubes can ensure reliable nutrition and facilitate easier medication administration when swallowing is difficult or stressful. While placement does involve a procedure and a small risk of infection, many families find that the benefits — more energy and steady growth — far outweigh the challenges.

To ensure a child is getting enough nutrition and hydration, regular check-ins with a pediatrician or a GI specialist are crucial. Growth charts, urine output, and guidance from a dietitian can help confirm whether a child's intake meets their needs, even when food choices feel limited.

Reflux can cause further irritation over time, which is why symptoms like chest discomfort, feeding refusal, or frequent vomiting shouldn't be ignored. An endoscopy may be recommended to evaluate for inflammation, and acid-blocking medication can protect the esophagus and maintain comfort.

Constipation is another frequent concern. A combination of stool-softening and stimulating medications often helps, along with simple routines such as sitting on the toilet after waking up or after a meal when the gut naturally wants to move. A small footstool during toilet time can make pushing easier. In rare or stubborn cases, doctors may consider treatments such as botulinum toxin (Botox) injections to relax the anal sphincter, allowing stool to pass more easily.

Sometimes constipation leads to what looks like diarrhea. A hard stool can act like a "cork," allowing only liquid stool to leak around it. In those cases, clean-outs or rectal treatments, such as suppositories, can help reset the system.

A colonoscopy is typically not necessary for constipation alone. It becomes more critical if a child is struggling with very slow growth, ongoing diarrhea, or blood in the stool—symptoms that may indicate inflammation.

Because many children with CdLS communicate discomfort through behavior, it's not always clear whether a change in eating or self-injury stems from pain or from a behavioral challenge. An upper endoscopy can help determine whether the issue originates from reflux, stomach irritation, or another underlying medical cause. More specialized tests, such as gastric emptying studies or esophageal manometry, are available when needed, however they are rarely the first step.

Diagnostic testing is always tailored to the child. One of the most common studies for CdLS is an upper GI series, which checks for intestinal malrotation. Abdominal X-rays are sometimes used when a physical exam is difficult, or when it's unclear whether diarrhea is tied to constipation.

Families play a powerful role. You're often the first to notice when something shifts, whether that's a change in appetite, mood, sleep, or bowel habits. Trust those observations. When you share your observations with the care team, it helps ensure that your child receives the support they need to grow, explore, and enjoy food in ways that feel safe and comfortable.