The Importance of GI Testing

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Most individuals with CdLS have some type of gastrointestinal (GI) involvement. Most commonly, this includes gastroesophageal reflux disease (GERD), in which the valve between the stomach and esophagus, the tube which connects the mouth with the stomach, does not function properly and allows the acidic stomach contents to get pushed up into the esophagus. Symptoms include frequent vomiting, bad breath, destruction of enamel on the back of the teeth and abdominal pain ("heartburn"). With persistent reflux, there can be damage from the acid on the wall of the esophagus, including ulcers, hiatal hernia or Barrett's esophagus, in which the lining of the esophagus changes to be more like the stomach. This latter complication can bear a risk for developing a type of cancer (adenocarcinoma) of the colon. GERD is treatable, either by medications or surgery, but can present at any age and recur, so it is important for all families to be aware of it.

In some patients, malrotation, or abnormal twisting and tethering of the small intestine, can be present at birth. If present, malrotation should be surgically repaired. If not repaired, it can lead to volvulus, an acute occurrence in which the bowel twists on its stalk, resulting in cutting off of blood supply to the intestine. This can be intermittent and resolved, or it can persist and be a life- threatening emergency, and a not infrequent cause of mortality in CdLS. This more commonly occurs in the duodenum of the small intestine, but can occur in the cecum, or the start of the large intestine. Symptoms consistent with possible intermittent obstruction include acute and significant abdominal pain, rigid (board-like) abdomen, and/or bright yellow vomiting. If these occur, volvulus should be suspected and the child should be brought to the emergency room and/or a surgeon should be contacted immediately. Possibly X-rays and/or a definitive upper GI testing series should be done. These studies are recommended even if the child is asymptomatic upon arrival but with a history of this presentation. Another significant complication in CdLS is the risk for bowel obstruction, either by volvulus, or impacted fecal material, or other unknown causes. Presentation is similar to volvulus.

There are other rarer GI complications and the most important aspect is for all parents to be aware that these can occur so signs and symptoms are not missed. The Foundation has GI experts on the Clinical Advisory Board, who can answer questions related to GI complaints.