Gastrointestinal (GI) Involvement and Individuals with CdLS

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Gastrointestinal (GI) involvement is very common in CdLS. For babies with CdLS, feeding problems are the initial type of GI involvement. There can be difficulty with the mechanics of feeding, as well as tolerating an appropriate volume. Often a feeding tube, either a nasogastric tube (through the nose into the stomach) early in life or a gastrostomy tube (through the abdominal skin into the stomach) when the child is older, is required to help maintain the appropriate weight and take in the correct calories and medications. Later, the tube may be able to be removed. Constipation can also occur at any age in CdLS, likely due to slow bowel motility. This can be treated with fiber and/or medications.

The most common GI complication in CdLS is gastroesophageal reflux, where the valve between the stomach and esophagus does not function properly and allows the acid from the stomach to back up into the esophagus. This can occur at any age. Symptoms include frequent vomiting, bad breath, destruction of enamel on the back of the teeth and abdominal pain. Diagnosis is made by either an upper GI series, in which x-rays are done as dye is swallowed, or an esophagogastricduodenoscopy (EGD), done under sedation, in which a tube is passed from the mouth down past the stomach to look for ulcerations or other physical signs and to take a biopsy. It is important to manage this appropriately, either by medications or surgically, since there is a risk for a type of cancer (adenocarcinoma) of the bowel if left untreated. Reflux can be a hidden source of pain in individuals with CdLS.

In about 10% of people with CdLS, malrotation, or an abnormal rotation and tethering of the small intestine when it was formed, can be present at birth. This can be detected by an upper GI series, following it through to the duodenum (first part of the small bowel). 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, thus cutting off blood supply to the intestines. Symptoms consistent with possible intermittent obstruction include acute and significant abdominal pain, rigid abdomen, and/or bright yellow vomiting. If these occur, they should be addressed immediately. 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 series should be done. Another significant complication with individuals with CdLS is the risk for bowel obstruction by volvulus, impacted fecal material, or other unknown causes.

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 (CAB), who can answer questions related to GI complaints.