



## Gastroenterology

An individual with CdLS should have regular evaluations and immunizations with the primary care provider.

### **Malrotation and volvulus**

- At time of diagnosis, an upper GI series should be done to rule out malrotation. If malrotation is detected, early repair may be indicated.
- Rarely, a patient who has had repair of malrotation (Ladd's procedure) may still be at risk of midgut volvulus.
- Patients with and without malrotation have been reported to present with volvulus or twisting of a part of the intestine, which can result in serious illness or even death without prompt intervention.
- Any sign of potential volvulus (e.g. bilious emesis/vomiting bile or bilious withdrawal from gastrostomy tube, sudden acute abdominal pain or distension) should merit an immediate visit to the emergency room, work-up and potential surgery.
- Ensure that the family has the CdLS Medical Alert Card, available from the CdLS Foundation Web site, which would be helpful in an emergency situation (e.g. risk for volvulus).

Support organization information should be given to the family whenever a diagnosis is made:

The CdLS Foundation

1-800-753-2357

[www.CdLSusa.org](http://www.CdLSusa.org).

## **Gastroesophageal reflux (GE reflux)**

- GE reflux is very common in CdLS.
- Many patients will present in infancy with signs of GE reflux including vomiting and irritability. In this case, empiric treatment is indicated with acid blockers +/- prokinetics.
- Some patients may have silent reflux, with a change in behavior as the only sign. Children with CdLS with undiagnosed GE reflux may present with self-injurious behaviors and outbursts.
- Evaluation for gastroesophageal reflux disease (GERD) may include pH probe/impedance study, nuclear medicine scan and/or endoscopy.
- With any clinical suspicion of worsening or initial signs of GERD, a repeat evaluation should be performed.
- Therapies include medications (acid blockers +/- prokinetics), post-pyloric feeding and surgical fundoplication (Nissen or Thal procedure). Management and treatment guidelines for GE reflux are available [www.cdlsusa.org](http://www.cdlsusa.org).

## **Feeding, growth and nutrition**

- Many children with CdLS have feeding difficulties in infancy, and some children are dependent on tube feedings throughout their lives.
- Use CdLS specific growth curves to follow weight, height and length.
- Children with CdLS are generally small, but should still grow along their own trajectory. Many children with CdLS require placement of a gastrostomy tube for maintenance of hydration and nutrition.
- Age appropriate formulas may be used. Some families prefer to use blended diets, which may be done safely under the supervision of a registered dietician.
- Aspiration is quite common in CdLS. Concern for swallowing difficulty or pneumonia should prompt further evaluation including a modified barium swallow.
- Manage constipation with fiber and/or medication as indicated.
- Whenever any surgery is performed, all involved specialists should be consulted in order to maximize the use of anesthesia and so that the individual can undergo diagnostic or management studies as needed at the same time.