

Loeys-Dietz syndrome

Gastrointestinal Issues

Many individuals with Loeys-Dietz syndrome (LDS) have gastrointestinal issues, often associated with food allergies. Because each person with LDS is different, it is important to talk with your doctor about what care is right for you.

What are the main gastrointestinal concerns for people with LDS?

- **Food allergies** - People may be allergic to one food or to many. A person can be allergic to any type of food, but frequent food allergies include milk, eggs, peanuts, tree nuts, fish, shellfish, soy and wheat. People with LDS are more likely to have food allergies than the general population. Symptoms range from mild to severe, and may include rashes, trouble breathing, abdominal pain, abnormal bowel movements, poor weight gain, reflux, vomiting, esophageal spasm, and decreased appetite. Food allergy responses range from chronic, low-grade symptoms to acute life-threatening reactions. People with food allergies are frequently prescribed injectable epinephrine (Epi-Pen) to use in emergencies. Instructions for epinephrine use are different for people with LDS than for the general population and should be discussed with your doctor.

- **Eosinophilic Gastrointestinal Disease (EGID)** - Eosinophilic esophagitis, gastroenteritis, and/or colitis - In some individuals, food allergies can result in allergic inflammation in the intestinal lining. When there is intestinal inflammation there are extra white blood cells in the lining of the intestine. These specific white blood cells, known as eosinophils, are active in allergies. You do not have to have other symptoms of food allergies to have EGID. People with LDS are more likely to have this kind of inflammation than the general population. This condition requires biopsies of the GI tract to diagnose. Symptoms of eosinophilic esophagitis (EE/EG) include severe heartburn, difficulty with swallowing, food impaction in the esophagus, nausea, vomiting, diarrhea, and weight loss.

- **Inflammatory bowel disease (IBD)** - A smaller subset of people with LDS have been diagnosed with IBD, a chronic inflammation of the intestines. IBD is characterized as either Crohn's disease or ulcerative colitis, depending on the site of inflammation. Crohn's disease can involve inflammation anywhere along the digestive tract, whereas ulcerative colitis is specific to the colon (large intestine). In both cases, inflammation of the intestinal walls causes bleeding and prevents proper absorption of nutrients.

Common symptoms include nausea, diarrhea, abdominal pain and bloody stools. These symptoms can often cause people to feel tired and lose their appetites and may be accompanied by joint inflammation, eye problems or anemia due to blood loss. IBD may cause growth problems or a delay in puberty because it can interfere with a person getting nutrients from the food they eat.

- **Constipation** – Many people with connective tissue disorders, including LDS, have constipation. When someone is constipated they can have large, hard, difficult to pass bowel movements, small pebble-like bowel movements, or even alternating hard and liquid stools. Severe constipation can lead to frequent accidents or leaking of stool between bowel movements.

How are gastrointestinal issues diagnosed?

The diagnosis and monitoring of gastrointestinal issues associated with LDS is based on a combination of exams:

- Laboratory tests - Blood tests can help identify the presence of allergies, and blood and stool tests can help detect inflammation and anemia.
- Endoscopy - Several types of endoscopy are used to diagnose IBD and how much of the intestines are affected. Endoscopy involves using a thin, flexible tube with a lighted camera inside the tip, which allows doctors to look at the lining of the gastrointestinal tract. The image is magnified and appears on a television screen. Endoscopies are also used to obtain biopsies (small samples of tissue from along the intestinal wall) for further studies. Each procedure is named for the part of the GI tract examined: Sigmoidoscopy (the lower third of the large intestine), colonoscopy (the large intestine), EGD (Esophagogastroduodenoscopy - the lining of the esophagus, stomach (gastro) and duodenum (first part of the small intestine)), or ERCP (Endoscopic retrograde cholangiopancreatography - the bile ducts in the liver and pancreas). There are additional concerns for someone with LDS undergoing endoscopy compared to the general population that should be discussed with your doctor.
- X-rays, CT, or MRI imaging

Diagnosing variations of intestinal inflammation is not always straightforward and it may be years until a patient receives a correct diagnosis. Even without a definite diagnosis, patients can be treated to reduce the likelihood of flare-ups and to allow the proper function of the gastrointestinal system.

What is the treatment and management of these gastrointestinal problems?

While some patients outgrow food allergies, many food allergy-related gastrointestinal problems, IBD, and EE tend to be chronic. Patients may be symptom-free for years, but generally require maintenance medications to reduce the likelihood of flare-ups. Doctors need to work closely with patients in devising a management strategy, which may involve dietary changes and medications and, in rare cases, tube feeding or surgery.

If lab results suggest food allergies, initial treatment is typically avoidance of the foods to which a person has a positive allergy test. Foods are then re-introduced gradually. When avoidance of allergic triggers does not successfully relieve symptoms, steroids may be used to halt the inflammatory process in the short-term, with tailored medications used for long-term management. In rare cases, feeding tubes may be necessary for children with LDS who are not able to get enough calories for growth because of their GI diagnosis and symptoms.

Many children may be prescribed injectable epinephrine (an Epi-Pen) to use when exposed to the allergen. Epi-pens release epinephrine, which works by constricting blood vessels to increase blood pressure, relaxing smooth muscles in the lungs to reduce wheezing and improve breathing, stimulating the heart (increasing heart rate), and working to reducing hives and swelling that may occur around the face and lips. The rapid constriction of blood vessels may be dangerous to an individual with LDS because of their underlying vascular condition. Thus it is important to discuss with your cardiologist and/or allergist when to use an Epi-Pen during an allergic reaction. If not a life-threatening emergency, use of Benadryl to control the allergic reaction may be recommended first.

To Learn More About Loey-Dietz syndrome

Read the other fact sheets on LDS. Find them online at the Loey-Dietz Syndrome Foundation Web site at www.loeydietz.org/resources.php.

