

Fortunately, there are lifestyle changes that can help reduce how often these symptoms happen or make them more manageable.

This fact sheet is designed to provide general information to help families understand how and why GI symptoms happen in Fabry disease. It also offers some suggestions that may be helpful in managing these symptoms and reducing their impact on quality of life. However, it's important to discuss GI symptoms and how best to manage them with a health-care professional (HCP) before making any lifestyle or treatment changes.

Want to find out more about Fabry disease?

Visit www.fabryfacts.com for more information about Fabry disease. Topics include genotypes, phenotypes, diagnosis, prognosis, and why gene variants (also known as mutations) matter in Fabry disease.

A guide for people living with Fabry disease

UNDERSTANDING AND MANAGING

Gastrointestinal symptoms

Gastrointestinal (GI) symptoms—problems related to the digestive system, such as abdominal pain, diarrhoea or nausea—are unfortunately very common in people living with Fabry disease. They are often among the first symptoms of the disease to appear, sometimes occurring in children aged 1 to 4 years and can become more severe with age. It's been reported that more than half of adults with Fabry disease suffer from one or more GI symptoms.¹

If you or a family member are living with Fabry disease, you may already be familiar with some of the GI symptoms the condition can cause.

References:

1. Zar-Kessler C, Karaa A, Sims KB, Clarke V, Kuo B. Understanding the gastrointestinal manifestations of Fabry disease: promoting prompt diagnosis. *Ther Adv Gastroenterol.* 2016;9(4):626–634.
2. Di Martino MT, Scionti F, Sestito S, et al. Genetic variants associated with gastrointestinal symptoms in Fabry disease. *Oncotarget.* 2016;7(52) 85895–85904.
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4. Mayo Clinic. Diarrhea/Diagnosis & treatment. Available at <https://www.mayoclinic.org/diseases-conditions/diarrhea/diagnosis-treatment/drc-20352246>. Accessed May 4, 2021.
5. Mayo Clinic. Nausea and vomiting/When to see a doctor. Available at <https://www.mayoclinic.org/symptoms/nausea/basics/when-to-see-doctor/sym-20050736>. Accessed May 4, 2021.

Please feel free to contact us with ideas and questions: patientadvocacy@amicusrx.com.

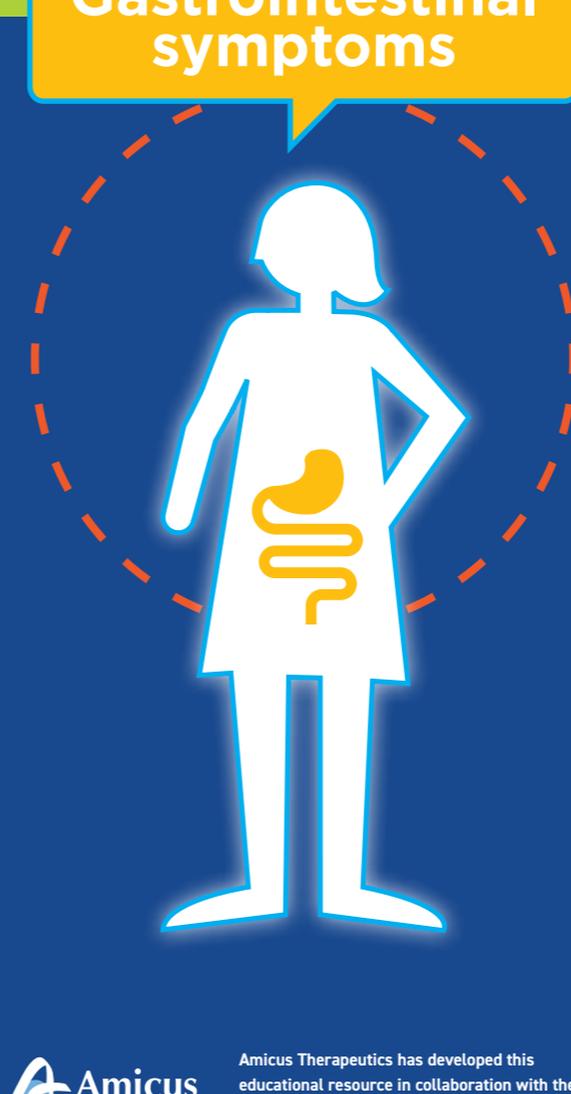


Please discuss any medical questions with a health-care professional (HCP). If you would like to provide feedback on this educational resource or would like additional information, please contact: patientadvocacy@amicusrx.com.

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What kinds of GI symptoms are associated with Fabry?

As is true of many Fabry disease symptoms, GI symptoms can vary greatly among people with Fabry disease, and manifest at different times in their lives. For example, some people may have frequent, severe abdominal pain starting in early childhood, while others may have almost no pain, but may frequently feel sick or have diarrhoea after meals, starting when they are teenagers. Similarly, some people may have one specific symptom that's very severe, whereas others may have several minor symptoms. GI symptoms also often change over time for many people living with Fabry disease.¹

The GI symptoms of Fabry disease can be very similar to those seen in other more common conditions. Also, Fabry disease is rare and may not be very familiar to some HCPs. Because of these two factors, Fabry disease is sometimes not recognised as the cause of GI problems until other Fabry symptoms arise later in the course of the disease. For example, Fabry disease-related GI symptoms may be mistaken for irritable bowel syndrome or other more common GI disorders. Unfortunately, this means that some people who have Fabry disease may live with these symptoms for a long time before they receive a correct diagnosis.¹

Why do people with Fabry disease often experience GI symptoms?

It's not known exactly why so many people with Fabry disease have GI problems.² As with other symptoms of Fabry disease, GI symptoms may be related to the accumulation of a fatty substance called globotriaosylceramide (or Gb3) in the cells. This accumulation is known to damage tissues and organs throughout the body. Some evidence suggests that damage to blood vessels and nerves in the digestive tract in particular, as well as damage to other tissues and organ systems, may play a role in the GI symptoms seen in people with Fabry disease.^{1,2}

Impact of GI symptoms

For some people with Fabry disease, GI symptoms can be far more than just an unpleasant inconvenience. When they are severe, these symptoms can significantly limit the activities people can enjoy with family and

Possible GI symptoms of Fabry disease

Abdominal pain	The most common GI symptom seen in Fabry disease is abdominal pain. Up to one-third of people with Fabry disease experience this symptom. People often describe it as a cramping sensation in the middle part of their abdomen. ¹
Diarrhoea (and/or constipation)	Diarrhoea (frequent loose bowel movements) is also very common. However, some people with Fabry disease may experience constipation (infrequent bowel movements that may be difficult or painful to pass). Others may have alternating periods of diarrhoea and constipation. ¹
Nausea and vomiting	Although it's somewhat less common, many people with Fabry disease experience nausea, as well as vomiting. ¹
Becoming full too quickly	Some people with Fabry disease start to feel full and lose their appetites after eating very small amounts of food. HCPs may call this problem "early satiety". ¹
Bloating	Some people with Fabry report feelings of bloating or gassiness in their stomachs or intestines. ^{1,2}
Heartburn	Heartburn—a burning sensation behind the breastbone and/or in the throat—may also occur. ²

friends, impair their performance at work or school and sometimes even keep them housebound. For example:

- People who suffer from frequent or urgent diarrhoea or vomiting may avoid going places where there may not be easy access to a restroom. Some may become reluctant to leave their homes at all.
- People who experience nausea or become full too quickly may eat too little, and not get enough calories and nutrients from their diets as a result.

Even if symptoms are not quite that severe, they can still have an impact on self-confidence and overall enjoyment of life.² It's no surprise that life-limiting medical problems (such as chronic GI symptoms) have been shown to increase the risk of clinical depression.³

What can be done about GI symptoms?

The most important step toward better management of GI symptoms is to talk to an HCP about them. Medications are available that may help reduce the severity of many of the GI symptoms Fabry disease can cause. Knowing more about the specific symptoms a person is experiencing can allow HCPs to provide treatments that are more likely to help.⁴

A symptom diary can be a great way to collect information about GI symptoms to share with an HCP. Keeping a symptom diary doesn't have to be complicated. Simply carrying a small notebook and writing down details about the symptoms as they occur (for example, when they happen, how bad they are, things that might have triggered them) can work well. Many free or low-cost apps are also available to help make it easy to track health-related information on a smart phone or other electronic device.

In addition, certain lifestyle modifications may help reduce GI symptoms. Some people may find that keeping a symptom diary also helps them identify—and then avoid—specific foods, activities or circumstances that provoke or worsen their symptoms. A list of lifestyle modifications that may help manage GI symptoms is provided in the chart to the right. Consider exploring them to see if they help.

However, be sure to talk to an HCP before making dietary or other changes.

Tips that may help reduce GI symptoms

Meal planning:



Eating smaller amounts of food more frequently may be helpful, especially if average meal sizes tend to worsen symptoms.¹

A lower-fat diet also may help make some GI symptoms less frequent and/or less severe.^{4,5}

Travel/going out:



It may help reduce anxiety to scout out the location of restrooms in unfamiliar public places in case of later need.

When travelling by air, keep medications in a carry-on bag. This will keep them close at hand and ensure that they won't go missing if checked luggage is lost or misplaced.

Stress reduction and rest:



Some people find that stress can make their GI symptoms worse.¹ In these cases, doing gentle yoga or learning how to meditate may help.

Getting enough sleep tends to help people feel their best overall, which may help reduce the impact of any GI symptoms they may experience.⁵

Staying hydrated



Keeping a refillable water bottle on hand can make it easier to stay hydrated. Small sips of cold, clear, carbonated or sour drinks such as ginger ale or lemonade can help relieve nausea and replace fluids lost by vomiting.^{4,5}

Cut off this chart and put it on your refrigerator to help keep these tips top of mind.

