

difficulties they can cause. But fortunately, there are some lifestyle changes you can try to see if they help you experience GI symptoms less often or make your symptoms more manageable.

This fact sheet is designed to provide general information that will help you understand how and why GI symptoms happen in Fabry disease. It also offers suggestions that may assist you in managing these symptoms and reducing the impact they have on your life. However, you should be sure to discuss your GI symptoms and how best to manage them with your healthcare provider (HCP) before making any changes to your habits or your treatment.

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 mutations matter in Fabry disease.

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.
3. Simon GE. Treating depression in patients with chronic disease. *West J Med*. 2001; 175(5): 292-293.
4. Mayo Clinic. Diarrhea/Preparing for your appointment [<http://www.mayoclinic.org/diseases-conditions/diarrhea/diagnosis-treatment/preparing-for-appointment/ptc-20233017>]; Diarrhea/Self-management [<http://www.mayoclinic.org/diseases-conditions/diarrhea/manage/ptc-20233046>]. Accessed September 26, 2017.
5. Mayo Clinic. Nausea and Vomiting/When to see a doctor. [<http://www.mayoclinic.org/symptoms/nausea/basics/when-to-see-doctor/sym-20050736>]. Accessed September 26, 2017.

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



Amicus Therapeutics, Inc.
1 Cedarbrook Drive,
Cranbury, NJ 08512
USA

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, diarrhea, 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 as young as 1-4 years old and can become more severe with age. It's been reported that more than half of adults with Fabry suffer from one or more GI symptoms.¹

If you're one of the people living with Fabry who struggles with GI symptoms, you are probably already familiar with some of the



What kinds of GI symptoms are associated with Fabry?

As is true of many Fabry 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 at all but may frequently feel sick or have diarrhea 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.¹

The GI symptoms of Fabry disease can be very similar to those seen in other more common conditions. Also, Fabry is a rare disease that may not be very familiar to some HCPs. Because of these two factors, Fabry disease is sometimes not recognized as the cause of GI problems until other Fabry symptoms arise later in the course of the disease. For example, Fabry-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 may live with these symptoms for a long time before they receive a correct diagnosis.¹

Why do some people with Fabry disease 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, 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.¹²

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 a third of people with Fabry experience this symptom. People often describe it as a cramping sensation in the middle part of their abdomen. ¹
Diarrhea (and/or constipation)	Diarrhea (frequent loose bowel movements) is also very common. However, some people with Fabry may experience constipation (infrequent bowel movements that may be difficult or painful to pass). Others may have alternating periods of diarrhea and constipation. ¹
Nausea and vomiting	Although it's somewhat less common, many people with Fabry also experience nausea, as well as vomiting. ¹
Becoming full too quickly	Some people with Fabry 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. ¹²
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 diarrhea 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 you do about GI symptoms?

The most important thing you can do to help manage your GI symptoms is to talk to your doctor or other HCP about them. Medications are available that may help reduce the severity of many of the GI symptoms Fabry can cause, and knowing more about the specific symptoms you experience can help your HCP provide treatments that are more likely to help you feel better.⁴

Keeping a diary of your symptoms can help you provide more detailed information to your HCP, and also may help you identify and avoid any specific foods, activities, or circumstances that trigger symptoms for you.⁴ Keeping a symptom diary doesn't have to be complicated—all you have to do is carry a small notebook and write down details of your symptoms as you experience them (for example, you can note exactly when you have them, how bad they are, and anything you suspect might have caused them). Or, if you prefer, you could try out one of the many available apps designed to help people track information about their health on a smart phone or other electronic device.

Some people may find that lifestyle modifications can help reduce their GI symptoms. To see if this is true for you, consider trying the suggestions listed in the chart to the right. (However, be sure to talk to your HCP before making any changes to your diet or other habits.)

Tips that may help reduce GI symptoms

Meal planning:



Eating smaller amounts of food more frequently may be helpful, especially if you often feel worse after regular meals or if you tend to become full too quickly.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 prevent problems and help you feel more secure to scout out restrooms in unfamiliar public places before/in case you need them.

When traveling by air, keep your medications in your carry-on bag. This will help ensure that you'll have them when you need them, even if your checked luggage is misplaced or lost.

Stress reduction and rest:



Some people find that stress makes their GI symptoms worse.¹ If this is true for you, doing gentle yoga or learning how to meditate may help.

Getting enough sleep is likely to make you feel better overall, which may help reduce the impact of any GI symptoms you experience.⁵

Staying hydrated



Keeping a full water bottle with you wherever you go can encourage you to stay hydrated. If you are nauseated or have been vomiting, small sips of cold, clear, carbonated or sour drinks such as ginger ale or lemonade may help you feel better.^{4,5}

Cutting off this chart and posting it on your refrigerator may help you remember to keep these tips in mind.

