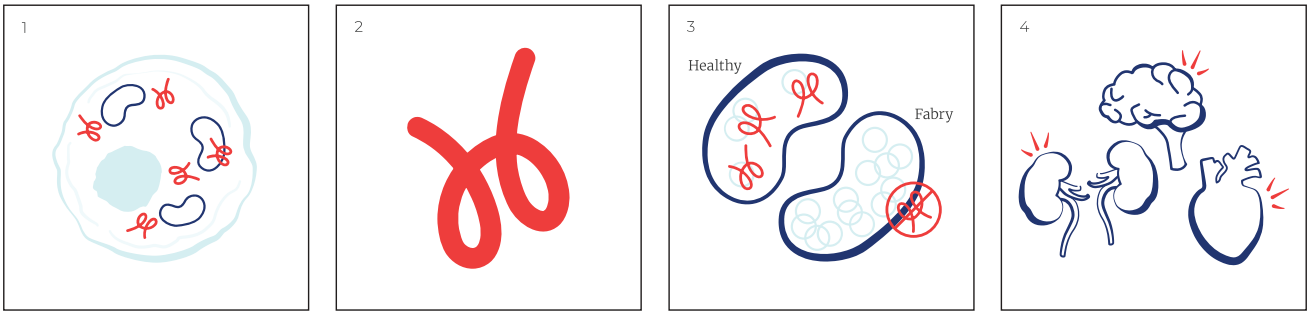
Questions your patients may have for you

Helping your patients understand what Fabry and its treatment means for them

Q: What is Fabry disease?

A: Fabry disease is an inherited condition.[1] It can affect different people in different ways, but it all starts with one thing: an enzyme.[1]

How the lack of one enzyme can lead to many issues



We all have enzymes inside our bodies to help our cells work.[2]

When someone has Fabry disease, their body can't produce enough working copies of an enzyme called alpha-Gal A.[1,3-5]

Normally, alpha-Gal A breaks down a type of fat that provides energy to the body.[4,5] But without alpha-Gal A doing its job, this fat can build up and damage cells throughout the body.[1,3-5]

Damaged cells lead to the signs and symptoms of Fabry disease.[1,3-5]

alpha-Gal A, alpha-galactosidase A.

Q: Does Fabry disease run in families?

A: Yes. Fabry is a genetic disease that is passed from parent to child. So when someone is diagnosed with Fabry disease, it's important for relatives who are blood-related to get tested.[6]

Q: What does Fabry disease look like at diagnosis—and how can it change?

A: Fabry disease can affect different people in different ways. For some, it may start with kidney issues. For others, changes in their heart rhythm will be the first indication of Fabry disease.[6] But, it's always a progressive disease. Fabry disease will not go away on its own, and you will likely experience more symptoms over time.[6]

Q: Can treatment help a progressive disease?

A: Yes. While there is no cure for Fabry disease, starting treatment early may slow it down.[7]

Q: Can Fabry disease affect women?

A: Yes. Fabry disease can also significantly affect women, with some women experiencing disease severity that's very similar to its impact on men.[8]

Q: Can both men and women pass Fabry disease on to their children?

A: Yes. Fabry disease is passed down by both men and women on the X chromosome.[9] Women with Fabry disease will have a 50% chance of passing the gene to their child regardless of its gender. Men cannot pass the gene on to their sons, but they pass Fabry disease to all of their daughters.[10] Work with a genetic counselor to learn about how Fabry disease can be passed on.[6]

Q: Why is genetic screening for Fabry disease so important?

A: Identifying people with Fabry disease can help more people get ahead of the progression of the disease.[6,11] A study found that once a person is diagnosed with Fabry disease, genetic testing can identify 5 or more family members who also have it.[6]

Q: Who can be a part of your care team to help you manage your Fabry disease?

A: Many different types of doctors may contribute to your care.

- **Primary care doctor:** Monitors your overall health. If you aren't sure which specialist to talk to, your primary care doctor will be able to help[1]
- **Geneticist:** Tests whether you or your family members have inherited the disease.[6] They can help counsel you and your family on how to manage this information
- **Neurologist:** Keeps track of your pain, sweating, and other symptoms related to your nervous system[6]
- **Nephrologist:** Sees how well your kidneys are working and measures levels of protein and vitamins in your urine and blood[6]
- **Cardiologist:** Monitors your blood pressure and any irregularities in your heart[6]
- **Gastroenterologist:** Checks for possible damage to your gastrointestinal (GI) system[6]
- **Other specialists:** Depending on your signs and symptoms and overall health,[6] additional healthcare professionals may be included on your care team

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