

SPEAKER: So the most common GI manifestations of Fabry disease are abdominal pain is typically the most common and then diarrhea is the second most common. Patients will complain of severe abdominal pain at random times throughout the day. Sometimes it's associated with meals. Sometimes it's associated with exercise. Sometimes it has absolutely no correlation with any type of trigger. The thought behind that is that maybe it's related to some stressors on the body. Either there's not enough blood flow or oxygen to the gut and therefore, they're developing this abdominal pain.

In terms of the diarrhea, patients typically describe watery, frequent loose stools. I've heard of patients who have frequent stool incontinence where they won't be able to make it to the restroom in time due to the urgency. There is typically not an inflammatory process that is associated with the diarrhea, therefore, you will not see blood in the stool. Typically, there will be no signs of inflammatory processes when you check the stool looking for ideologies.

Other common symptoms that Fabry patients describe of their gastrointestinal manifestations are nausea and early satiety. Patients will say that they're eating and they won't be able to complete full meals or that they have nausea throughout the day.

Many of these symptoms are non-specific. We can see these in multiple other types of disease manifestations, and that's what leads to difficulty in making the diagnosis of Fabry disease based on gastrointestinal symptoms. Patients who have abdominal pain, who have diarrhea, who have nausea, and other symptoms, could also have something that is similar to irritable bowel syndrome, which is frequently a diagnosis of exclusion.

When patients present to the office with these symptoms, it's really important for clinicians, particularly gastroenterologists, to ask further questions about the disease presentation. There is nothing specific and there are no specific tests that will distinguish these gastrointestinal manifestations from other types of ideologies, but in addition to the patient's other manifestations of their disease and also family history. So it's important for the clinician to ask about specific other symptoms that the patient may be having that are not related to gastrointestinal symptoms. Including skin presentation and the neurological presentations.

We also frequently ask patients about their family history. If there is a cardiac history, if anyone in the family has had renal transplants, this will lead us to be more concerned about some genetic cause such as Fabry disease, and lead the provider to do further testing to look for Fabry manifestations.