

SPEAKER: When a patient with Fabry g.I. Symptoms typically presents to the doctor, the differential could include multiple different gastrointestinal diseases, including, as mentioned, irritable bowel syndrome. However, there's also the thought of other disease processes, including inflammatory bowel diseases such as Crohn's disease or all ulcerative colitis. Frequently, patients will also be treated for reflux disease, as it's thought to cause sometimes some of the symptoms of the nausea and the abdominal pain.

Other thoughts are celiac disease, which these patients are frequently screened for. However, when all of these tests and screening are normal and they have-- continue to have symptoms, these symptom-- these patients typically will be treated for an irritable bowel syndrome before they actually make the diagnosis of Fabry disease.

In a study that came out a while ago, they looked at patients who had GI manifestations and found that there is a delay of diagnosis of up to 10 years because these symptoms are so non-specific. And the practitioner will frequently just treat them as a functional abdominal patient or an irritable bowel syndrome type of patient without looking for other causes. That's why it's very important for the provider to ask the patient about their other symptoms that they're having, including any numbness or tingling in their hands and feet, any rashes on their skin, or any other symptoms that may be abnormal for them. Additionally, it's also important for the practitioner to ask about any family history that may lead them to think that there might be some genetic basis to the patient's symptoms.

As for adults, the patients also can present with new onset of gastrointestinal symptoms in their 20s and 30s or later, or they can be patients who never was seen by a gastrointestinal doctor in childhood and then present when they're older, although they've had symptoms for many, many years. These patients can have similar symptoms, including the abdominal pain or the diarrhea.

One of the interesting facts that I found with seeing patients is that a lot of the patients will have similar symptoms as their family members, including their parents and their siblings, their aunts and uncles and their cousins. And therefore, for them, these symptoms aren't that debilitating for them and don't cause as much concern. So I've had patients who show up with what I would describe as very debilitating symptoms, and they would not have seen a doctor about it. They continue to just function in their life and don't care that they have these symptoms because it seems that everyone does surrounding them and they think of this as a normal symptom. So this can cause a delay in being worked up in their symptoms because they don't seek care for their symptom presentation.

Finally, many patients also have described having severe abdominal pain, or diarrhea in childhood and early adulthood. And that actually improves as they get older without any other type of intervention. And the thought behind this might be that there might be some nerve burn out because of progression of the disease as patients get older. And so, therefore, the pain that they were experiencing is no longer because these nerves are no longer functioning. This hasn't been shown specifically in studies yet, but it's one of the theories behind the progression of the gastrointestinal manifestations.