

SPEAKER: Though, us in the Fabry community truly believe that GI symptoms are very important and are one of the initial presenting signs of Fabry disease and should be explored frequently as young patients. The bigger issue is that the incidence of Fabry disease is extremely low from the random population when there is no family history. And so it makes it very hard to convince providers to screen for Fabry disease, when the majority of patients who present with the exact same symptoms have an irritable bowel syndrome type of picture that is not a genetically based.

And so if you have 50% of the patients who are presenting with abdominal pain and diarrhea have irritable bowel syndrome and 0.01% have Fabry disease, it's hard for providers to do a screening for Fabry disease on all those patients who don't have it. I think it's more of an awareness of it that we need to make sure that providers know about so that they can ask for the associated symptoms and the family history to pick up on these patients so that they are treated early. And I agree that the whole push behind it is that we would love to start patients, particularly with no family history and no known Fabry in the family, on treatment as early as possible to prevent the progression of the GI symptoms.

In terms of patients who are initially diagnosed with Fabry disease based only on their gastrointestinal symptoms is extremely rare and almost unheard of at this point because the symptoms are so non-specific. And so the majority of patients who I have seen, even with severe symptoms, will typically have someone else in the family who went through a kidney transplant, or had a heart attack, or a stroke that led a provider to think more about the diagnosis of Fabry disease.