SPEAKER: GI symptomatology, male versus female-- there've been multiple studies that have tried to look at the differences between the sexes. However, repeated studies have shown that they're actually fairly similar. Men tend to present earlier with the GI symptoms, and they tend to be more severe earlier on compared to their female counterparts. However, as disease progresses, they both-- both males and females reach the similar levels of severity and percentage of patients having them-- having GI manifestations. And so there doesn't seem to be a significant difference.

Studies do show, though, that women tend to have more constipation than men do. However, this is also seen in the greater population of non-Fabry patients, where women tend to have more constipation than men. So this may just be more women than men in general as opposed to specific to Fabry Disease.

So the gastrointestinal symptoms of Fabry Disease can cause significant distress and morbidity for patients. And it's very important for it to be assessed and treated as soon as possible. Patients can present at any age with gastrointestinal symptoms. Although it is one of the initial presentations in Fabry Disease in younger children, adults can also have new onset of gastrointestinal symptoms, and they could also have gastrointestinal symptoms for decades that has never been evaluated or treated.

So as a provider, it's important to always ask about these symptoms and to look further into the etiology of them. These symptoms include abdominal pain, diarrhea, nausea, early satiety, and symptoms of reflux too. As a provider, if a patient presents with any of these symptoms at any age, it's important to ask more specifically about the symptoms, their age of onset, and their progress over the years or progression.

Additionally, in order to diagnose these patients, as these symptoms can be very non-specific, it's really important to ask the patient about additional symptoms that might be correlated with their GI presentation, including any signs of neuropathic issues, such as numbness or tingling in the hands and feet, any skin manifestations, or any other issues that might be related to Fabry Disease. Additionally in these patients, it's always important to think about a family history, as this will lead one to consider a genetic cause to their symptoms. So if a patient presents with symptoms and has a family history, a brother, sister, parent who had similar symptoms or has early onset of renal, or cardiac disease, or stroke, always think about a genetic cause such as Fabry Disease that might be leading to their GI symptoms.

As gastrointestinal symptoms can be the initial presentation or early presentation of Fabry Disease and studies have shown that early treatment of Fabry Disease can lead to improvement in these symptoms and in symptoms and other systemic parts of the body, it's very important for these patients to be diagnosed early. Therefore, as a provider, it's important to always have a suspicion for Fabry Disease when a patient presents to your office. If this disease that's picked up early and treatment is targeted for the disease process, these symptoms can improve significantly and also prevent worsening symptoms in the future.