

SPEAKER: First, to start off with, there are some states now that have Fabry disease as part of the newborn screen, which we'll pick up on some of those patients with mutations. Beyond that, then the screening is in general pediatric offices, and then in sub-specialists when patients present.

In the gastrointestinal office, if a patient is presenting with any of these signs or symptoms, it is important to ask about the specific family history. So if anyone else in the family has similar disease presentation, as there is a thought within Fabry providers and researchers that there may be a GI phenotype of Fabry disease, and that one of the mutations or phenotypic presentations is more severe GI disease. And so if there's someone else in the family or multiple people on the failing have similar presentations of severe GI symptoms, this will lead to more of a workup for a genetic basis to it.

Additionally, if there seems to be people in the family who have early onset of renal disease that haven't been diagnosed, or cardiac disease, or vascular disease, those are important questions to ask the patient and the family. And so whenever a patient presents with non-specific symptoms, it's really important to get more of a general family history.

In terms of screening, the screening is typically done if there's a suspicion beyond just the presentation of their symptoms. So either if the patient has additional comorbidities or if there's the family history that would warrant further screening for Fabry disease that should be undertaken. If there is a positive diagnosis of Fabry disease, then there's a further discussion about screening others in the family.