SPEAKER: One of the questions that we get frequently about Fabry disease is how do we keep from missing cases? And how do we increase our sensitivity? There are a few strategies for that, and they're going to differ a little bit between the classical Fabry disease and the non-classical.

So for a classical Fabry disease, we often get a delay in diagnosis. So the earliest symptoms are commonly in childhood, typically for boys around age five or six, for girls around 9 or 10. But the diagnosis is often not made until early adulthood. So then the reason for that is that the symptoms-- the early symptoms are recognized retrospectively, but the early symptoms are quite non-specific.

So limb pain is a common pediatric complaint. And it doesn't trigger an aggressive workup for an underlying diagnosis because it's generally multifactorial, non-specific complaint. Same thing with GI symptoms, and those are the main symptoms that show up.

Now, you can get some clues in classical Fabry disease by asking about decreased sweating, early fatigue, and by getting a family history. The family history is probably your most valuable tool for pediatric diagnosis. And a good eye exam-- but it has to be with a slit lamp-- can also be a valuable tool. So if you have somebody who you think maybe has Fabry disease and you don't order a lot of genetic testing, if you refer to a pediatric opthalmologist, almost all of classical patients will have corneal whirls, so you can use that as a screening evaluation before you would order a genetic test.

For adults, you see delays in treatment just because the disease is rare. Classical fabry disease is about 1 in 40,000. And that isn't going to be at the tip of people's tongues. So how do we get people to think of it? One is just by talking about Fabry disease more openly. The advent of therapy for Fabry disease has really improved awareness of the disease dramatically, but we still do have adults coming in with classical Fabry disease where that question has just never been answered.

Some of the things that have been recommended for screening adults are anybody with proteinuria or with unexplained progressive kidney disease. And probably all patients with hypertrophic cardiomyopathy should be considered for screening for Fabry disease.