SPEAKER 1: When I'm thinking about Fabry Disease and how it presents, I organize it in the following way. You have the medical student presentation, which is the easiest to recognize. The most common initial presentation for a new patient with Fabry Disease is, I have a family history of Fabry Disease. And that was something that nobody should miss.

The second things that come along that are also fairly recognizable are if somebody has corneal whirls or angiokeratomas, which are purple spots on the skin. In Fabry Disease they tend to cluster around the belly button and in the groin area, but can also occur on the lips, on the palms of the hands, and other areas of the body. Those are strongly associated with Fabry Disease and rare in most other conditions, although they can occur with some other conditions.

The presentation of family history of Fabry Disease, angiokeratomas, or corneal changes that are suggestive of Fabry Disease are quite specific, and should just automatically lead to questioning Fabry Disease and testing for it.

The next level is sort of the master clinician level. And that is if you get a presentation with proteinuria, cardiac rhythm disturbance, or hypertrophic cardiomyopathy. And you ask follow-up questions and then think could this be Fabry Disease and pursue that. If the diagnosis ends up being Fabry Disease, people will think, wow, that person's really good.

To pick up the other symptoms, which include GI pain, chronic recurrent diarrhea, burning pain in the hands and feet, ringing in the ears, hearing loss, and other non-specific findings you'd really have to be good. Or have to be somebody whose practice focuses on rare diseases, to use those as the basis for doing testing for Fabry Disease.