SPEAKER: GI symptoms are one of the most common and early presenting symptoms of Fabry disease. However, the GI symptoms are usually not one of the key diagnostic findings that leads to testing for alpha-galactosidase deficiency. The reason for that is that GI symptoms are fairly common in the general population, so having abdominal pain is not at all unusual as a pediatric complaint or in adulthood, which means that when people come in and complain of GI symptoms, Fabry disease often doesn't come to the top of the list of considerations very quickly.

If somebody wants to explore that, there are a few simple questions that will often help to confirm a suspicion or to raise the idea of potential Fabry disease. If GI symptoms are accompanied by burning pain in the hands and feet, by a family history of kidney disease, or a personal history of proteinuria, or of cardiac rhythm disturbance, or hypertrophic cardiomyopathy, then Fabry disease becomes much more likely.