SPEAKER: So many of you may face the challenge of deciding which patient may be a target for further testing to identify Fabry as the underlying pathology. And so I'll tell you that, sometimes, that can be a challenge, especially in a clinic where you're very busy and seeing a lot of patients.

So a few of the things that might help, that we would try and do in cardiology in general is to ask about unexplained death or cardiovascular death before the age of 50. And if you hear that, always be concerned there may be a genetic driver for that in the family. Also early cerebrovascular accidents are very strongly associated historically with Fabry Disease.

But the things I would tell you are the way that I approach patients that come to me with a concern of hypertrophic cardiomyopathy. So for me, all of those patients get genotyped. And I recognize that may not be the practice that all of you pursue, but let me explain why that may be advantageous.

So one is that you can do that screening, and those panels are going to include traditional, quote unquote, "causes of classic hypertrophic cardiomyopathy" such as sarcomeric mutations. But they'll also have some diseases on there that have implications outside of the cardiovascular system. So that might be amyloid, which you're reading more and more about every day. That might be Noonan syndrome. That might be Fabry.

And so we've worked with these companies to develop these panels to capture not only the typical causes but some of these more nuanced causes or rare causes of disorder. And so, many times, I'll find out about Fabry because of a genetic panel that we've performed and we see that hit that helps us to look a little bit more into the possibility of Fabry.

In our clinic, we have a little more time because it's sort of a sub-subspecialty clinic multidisciplinary approach where we have the time to ask about things such as problems with sweating, problems with pain, problems with school absences, from GI discomfort, all these other sorts of things.

And I recognize, in clinical practice, that's probably just not going to happen, especially in adult cardiology. We're not going to go back and spend a lot of time talking about your childhood and your adolescence. But you should be pursuing a family history. What I would say, though, is to think more about potentially leveraging genetic testing in your patients with a hypertrophic phenotype.

We can talk about the other cardiomyopathies, but HCM is a nice target because we know many of the genes that are implicated in hypertrophic cardiomyopathy. Obviously, we know the genes for Fabry. We know many of them for Noonan, other things like that. And that's going to give you an opportunity to learn a lot about these patients that, otherwise, you're not going to learn from an MRI, you're not going to learn from an echocardiogram

And the reason to do this is important. So as we've talked about in other segments, certain diseases such as Fabry have therapeutic options outside of AHA/ACC guidelines. You're not going to see statements about enzyme replacement therapy in the heart failure updates.

So it's important to understand that we're going to apply the traditional therapies. If they're hypertroph, we're going to monitor for arrhythmias and look for a potential for AFib and those sorts of things. If they have a dilated cardiomyopathy, we're potentially going to throw at them ACEs or ARBs or ARNIs and beta blockers and all those guideline-based therapies.

But they differentiate themselves because they have an additional therapy that you can offer, which would be potentially chaperone therapy, might be enzyme replacement therapy. The other reason you want to know about Fabry is, let's say they come to you purely for a cardiac complaint and you identify that patient has Fabry.

The other things you should do is work them into the appropriate providers that are outside of the cardiovascular system. So meaning, they need to see a geneticist. They may need to see a nephrologists. They may need to see an ophthalmologist. They may need an orthopedist or a pain management doctor. So this is very different than just dealing with that patient with isolated myocardial disease.

This is a systemic disorder, and so identifying it has implications for the patient and their treatment, but it also has implications as treating the patient as a whole, meaning we're not just worried about the cardiovascular system. We want to make sure the kidneys are doing OK. We want to make sure the brain is doing OK. So all these things have to be considered, and doing genetic testing will open that door.

So I would suggest to you that, in these patients, you have concern of some genetic trigger, meaning they have hypertrophy and it's not in the setting of uncontrolled systemic hypertension, for example. Consider genetic testing. And if you do, you may find someone with Fabry. You will find someone with Fabry if you do it enough, and that's going to make a difference in that patient's outcome. So I would urge you to learn more about that. And if you would like to learn more about how to pursue those tests, please feel free to reach out to me.