

SPEAKER: As you may have appreciated from some of the other modules, identification of Fabry can sometimes be challenging. As a clinician, you see many patients every day, whether you're in the pediatric realm, or in the adult realm, or in both, and so consideration of Fabry probably won't always be at the top of your differential.

I would urge you to think about it in patients as we talked about that have hypertrophy that is unexplained that you may think is a genetic trigger. And I think as a provider one of the best ways to understand more about genetically-triggered diseases, whether those are vascular, or cardiomyopathy, or channel-based diseases, is to take a family history.

And there are programs out there that can be done that make it quite efficient from a time perspective that can be done by someone other than a health care provider, such as a cardiologist. So potentially a nurse or an MA may be able to help you with that. But if you start seeing patterns on that pedigree, you should automatically think about potentially investigating for heritable causes of cardiovascular disease.

And then as much time as you can devote to patients to get a more thorough and broad history is always advantageous. So for those patients that you see that have a hypertrophy ventricle, perhaps stop and ask you, do you have problems with sweating? Have you had problems with GI complaints? Is your kidney function abnormal? And all these things will be clues to point you in the right direction.

I still think that many of the opportunities to diagnose Fabry will come through genetic testing, whether that's at the neonate level or whether that's in your clinic when patients come to see you, and you do this panel as a workup. And as I said, it's an important thing for people to know, because it changes your management strategy. It's just that simple.

And for those patients out there that are suffering from HFpEF, for example, as you know, we don't have a lot of great therapeutics. So if you had something to offer at least a few of those patients, that's an important opportunity, and I want to urge you to take that.

I think the biggest target for all of us is to increase awareness of this disease. So many of the patients that I follow are self-referred. They find out about the diagnosis and they start googling and looking for things. And they find a few providers, many of which you may have listened to throughout this series, that have recognized expertise in the area of Fabry.

And you have to remember, it's still not a very common disease. Obviously, the prevalence is probably a low estimate, because we don't screen everyone, so we don't know how many patients there really are with Fabry. But there are more than what we have, I'm confident about that.

But that one Fabry patient in a rural area is likely going to be the only Fabry patient that that cardiology group follows, or in your internal medicine clinic. And so what I advocate is that whenever possible, you should have these patients seen at a center of excellence, where they can get a nice overview and plan for their cardiovascular care, for their kidney care, for all these other sorts of things, recognizing there's some patients that can't travel.

And that's why I would urge you to take advantage of these opportunities of the speakers that you hear from, to reach out to them, and to see if they can offer you guidance. And I do this very frequently and enjoy being able to do it. I like being able to help people, even if they can't come and see us in person.

But I think as a group, whether that's as cardiologists, whether that's as providers, whether that's as physicians, we do need to make a stronger stand in increasing awareness through social media, through guidelines, all these other sorts of things that need to be augmented to make sure people are aware.

And it's amazing how many people will come in and say, you know, once I started reading about this disease, it explains my history to a t, from when I was five years old all the way to an adult. And so it's a serious disease. It has major impacts on quality of life. It has important implications on the cardiovascular and the renal side.

And so awareness is always a good thing, which leads to screening and looking for these patients. And I think that first order is going to be genetic testing. And then you start to cascade through the other possibilities about looking in a thoughtful way at the cardiovascular system and the right kind of testing over time.

And we are starting to accrue more and more data. Fabry is becoming a much more discussed topic in cardiovascular circles, especially, I think, in part because of the advent of genetic testing and because of some of the newer technologies that we have are available, such as CRISPR Cas9 technology.

So those things are starting to really increase some of the momentum behind care for Fabry Disease. So this is a time for us to become more thoughtful in how we do it. We do have registry data. But as many of you know, registries typically have many holes, where all the data were not collected. Maybe the data aren't accurate sometimes.

And then as we do different tests, we don't have a database of those findings, meaning I don't have 20 years of data worth of cardiac MRI, because we haven't been doing it that long. So there are certain limitations to all this, but the one way to overcome it is to capture as many patients as we possibly can and to cohort all of that information in one place so we could potentially look at that at least for outcomes research.

So I urge you to consider Fabry in your practice. If there are any questions that can ever be answered about cardiovascular implications, please feel free to reach out to me.