

SPEAKER:

Now, for non-classical Fabry disease, usually you will not have pediatric symptoms. And a lot of times, the symptoms will relate only to one system of the body. So you may have what appears to be isolated kidney disease or isolated heart disease. And really, for that, the bottom line is you have to have a low threshold for when to test. And again, every patient with unexplained progressive kidney disease warrants an evaluation to see what the underlying condition is, as does every patient with a progressive arrhythmia or with hypertrophic cardiomyopathy.

So one of the strategies that I think should be implemented-- currently, we know that hypertrophic cardiomyopathy is often genetic, and there are panels that have genes that cause hypertrophic cardiomyopathy. The hypertrophic changes when you look at the heart itself using echo or cardiac MRI are going to be similar if it's Fabry disease versus some other form of heart disease. I think that we should make it so that the panels that look at causes of hypertrophic changes in the heart include both the sarcomeric proteins, which is the most common form of hypertrophic cardiomyopathy, and the alpha-galactosidase A deficiency, which causes Fabry disease. That will increase the recognition and allow for the most specific treatment to be implemented in a timely fashion.

A similar strategy could be used for suspected kidney disease. If there is a reason to do a genetic test, and we say, OK, everything that causes proteinuria should be included in the testing algorithm. That will result in improved diagnosis, not only of Fabry disease, but also of other genetic kidney diseases. So I think in the adult population, that strategy would make a lot of sense.

As far as just general awareness of Fabry disease, just knowing what the symptoms are and having a low threshold for testing it-- you don't have to have a hit every time you order the test. In fact, if you are right every time you order the test, you're probably missing a lot of cases and need to test more frequently.