SPEAKER: So there is an obvious interest in therapeutics in the area of Fabry Disease, and you've heard about some of them through these different modules. Many patients and families have heard about enzyme replacement therapy, which is available in the United States and obviously available across the globe.

There are other therapies such as chaperone therapy, which works in patients with admittable mutations, and you can find more information about this in other modules. And then this idea of editing the genome. Could we potentially use technologies to replace diseased DNA segments or genes with healthy copies? And all of these are actively being assessed.

There are alternative enzyme replacement therapies that are being studied as we speak looking at different things than traditional enzyme replacement, so meaning for enzyme replacement to potentially be more efficacious, perhaps if you had more of that activity that was detectable in the blood for a longer period of time, in theory, it may work better.

If, perhaps, from a quality of life, you didn't have to give the therapy as frequently, that might impact decision-making for patients. And so all these things are very important, and all coming down the pike.

And it's actually somewhat beneficial for Fabry, because for disorders that are single-gene disorders, it's a much easier target than for me to go after systemic hypertension, which may be polygenic, and environmental, and other things that have implications. So Fabry makes this a reasonable target to look for these sorts of interventions.

The problem is that we don't have lots and lots of patients that we can put into trials, such as RCTs. We'd probably never accrue a number of patients necessary to find significant power to do a longitudinal prospective study. And in the current milieu, I think it would be very hard to argue to do anything that's placebo-controlled, because we have pretty good data that conventional ERT does make a difference.

However, we do accrue data over time on all the patients that we can capture with Fabry through the Fabry Registry. And that's provided longitudinal data about outcomes and phenotypic implications. And so we can use existing data as a comparator, if you will, for these upcoming therapies.

And that's an important thing. It's better than just postulating that it makes a difference without any obvious endpoint to look at. However, we have to remember that as we've discussed in other modules, the way that we screen and treat patients is not the same today as it was in 2010. So using those historical controls inherently has its own limitations. But it at least gives us an idea if we're going in the right direction.

And I think the more that I take care of patients that have genetically triggered disorders, it's great to have options. And to only have one or two sometimes becomes a bit of a problem. And you can have situations for if you're receiving enzyme replacement that perhaps you develop some immunogenicity to that therapy, and you can't receive it as safely as you might like.

Or that perhaps it's a cost issue. Perhaps it's a quality of life issue. So to continue to look for these things is paramount, because we do think we can make a difference in the outcomes for patients. And even if we can only impact their pain, or if maybe I can reduce some of their scar burn on MRI, to me, that's worth the intervention.

And so this is a hotbed of intervention. So for you as providers and your patients, tremendous opportunities now that have not existed ever in the world of Fabry to enroll patients into clinical investigations. And I will provide my contact information. I would be happy to facilitate that for you, because this offers patients to have access to technologies that otherwise they won't.