DR. HEATHER A. LAU: Fabry disease is an X-linked lysosomal storage disorder that's due to a deficiency of alpha-galactosidase enzyme. When this enzyme is deficient, it leads to accumulation of a substrate called Gb3 for short.

Classical Fabry disease manifests early in childhood with a variety of symptoms. However, oftentimes, the initial symptom is neurologic. And they're in the form of painful acroparesthesias. The neurologic manifestations of Fabry disease range from peripheral nervous system involvement all the way through the central nervous system involvement.

If you look at a retrospective cohort of pediatric patients, 68% to 80% of them will present with pain as their initial symptom. This is often misdiagnosed or just completely ignored by their parents or practitioners. However, when severe, it can lead to painful crises that limit their exercise ability, their ability to participate in sports due to the other symptoms of classical Fabry disease which include difficulties with sweat or hypohidrosis. This will also limit their ability to participate in sports and activities.

What happens during a painful crisis is that a trigger such as heat or fever or exercise will lead to a particular pain that starts in the hands and feet, and can radiate upward. Most patients described this pain as chronic and burning pain. Others will describe it as shooting. The painful crises will be even more severe and require the patient to stop from what they are doing and recover.

Since Fabry disease is X-linked, most often, men are considered to be more affected, and they will have earlier onset of symptoms in the classical form of Fabry disease. However, females are affected as well. And even occurring in childhood with manifestation of painful crises and exercise intolerance from hypohidrosis. It's important that we not miss these young females because they, too, can benefit from therapy.