SPEAKER: Although Fabry is a multi-systemic disorder, the first signs of classical Fabry are pain and GI symptoms, which are non-specific. It leads to a long diagnostic journey because the other symptoms involving renal and cardiac come later in adolescence and adulthood. It is because of this that many children and adolescents are misdiagnosed for a long time.

For example, in one case of a 26-year-old man, who was diagnosed because his cousin was finally diagnosed with Fabry disease. And when I first met this patient, he described that for over 10 years, he had suffered from pain in his arms and his legs, specifically in his hands and feet. And it was discounted by his doctor and family.

It would be triggered by exercise, so he chose more sedentary activities at school. It led to an increase in his depression, and he also suffered from ADHD. It wasn't until his diagnosis at age 26 that he finally felt validated and that he understood what was going on with his body.

He then started on an oral chaperone therapy, and within five years, his painful acroparesthesias resolved. In addition, his non-specific GI issues, which he just attributed to having a delicate sub-- delicate stomach, or he would avoid certain foods, or make sure that he was near a bathroom-- all of those symptoms that he never actually voiced to a doctor even after his Fabry diagnosis also resolved while on oral chaperone therapy. He was caught before he had significant renal or cardiac involvement, and he remained stable on oral chaperone therapy for the ensuing years.

So this case highlights the importance of early diagnosis and how pain in a child in childhood may not lead to the appropriate diagnosis. So this is why it's important to understand that the initial symptoms of Fabry disease are not going to be renal and cardiac, but, in fact, are neurologic and gastrointestinal.