SPEAKER: If a patient presents with a symmetric painful sensory neuropathy, it is important for the neurologist to actually do a complete physical exam. They might obtain other clues to the diagnosis outside of the neurologic examination. So in addition to screening for some cranial nerve dysfunction, episodes that are suggestive of TIAs or stroke, they should actually look to the heart, to the skin, looking for angiokeratomas.

And if they can, ask about eye exams. Has anyone picked up any abnormalities in their cornea? These are all other signs and symptoms that could lead to the diagnosis of Fabry disease. And if a neurologist suspects that this painful acroparesthesia could be Fabry disease, they themselves can try to send testing, or they can simply refer to a clinical geneticist.

For males that are suspected of Fabry disease, a simple biochemical test looking at the enzyme activity can be sent. However, for females, actually, the confirmation of diagnosis must come from actual DNA testing. And this is because of X-- of its X-linked inheritance leading to X inactivation. Their enzyme levels may not be indicative of an affected person. So therefore, DNA confirmation must be obtained. And this can be done either through the neurologist if they're comfortable about their pre-test counseling as well as the method of testing, or they can simply refer to a clinical geneticist with their suspicions.