

SPEAKER: So in classical Fabry Disease, one of the earliest symptoms of the disease is pain. And this is pain typically localized to the hands and feet. The pain can be described by patients as chronic and burning or shooting. There are triggers to painful crises that include fever, exercise, and heat.

In addition to these painful crises or these acroparesthesias, the clinician should look for other signs and symptoms of classical Fabry Disease which have onset in childhood. This includes a variety of GI or gastrointestinal symptoms. They could be described as alternating between diarrhea, constipation, bloating.

And in fact, part of this GI dysmotility is due to neurologic dysfunction from Fabry Disease. In addition to those two signs, you can start to see angiokeratomas as well as cornea verticillata to help with the diagnosis of classical Fabry Disease. The diagnosis can be delayed if these other symptoms are not currently observed.

However, the clinician should remain suspicious when these painful acroparesthesias recur, and tend to have an association with a trigger. Both men and women can have painful acroparesthesias. Women tend to have a slightly later onset due to the nature of the inheritance of this disorder.