

SPEAKER: It is important for the clinician to recognize the neurologic manifestations of Fabry disease. It affects both the peripheral nervous system and the central nervous system. At times, it is the first symptom of Fabry disease in childhood or adolescence, and therefore, it's an opportunity to diagnose and intervene early to prevent the long-term progression of this disease.

The peripheral nervous system manifestations, as we described, include those painful acroparesthesias, but also include issues with sweating abnormalities, and even a component of GI dysmotility. There there's also autonomic dysfunction that can happen as well.

For older patients in their 30s and 40s, the other manifestations associated with neurologic dysfunction in Fabry disease include stroke and white matter burden. They may not be diagnosed as Fabry disease if they simply present with a stroke unless the clinician is looking for other symptoms or this is on their differential. For white matter burden, which is often misdiagnosed as either multiple sclerosis or microvascular disease from other cardiovascular risk factors, this is also often misdiagnosed and a missed opportunity to intervene and initiate treatment early.