

SPEAKER: The question that keeps presenting is at what point do you start treating patients with Fabry disease either with enzyme replacement therapy or chaperone therapy to try to treat the underlying disease? The thought at this point in relation to gastrointestinal symptoms is that many of the symptoms are presenting because of progressive accumulation of Gb3, leading to damage both in the vascular system, and in the nerves, and in signaling systems. The thought behind that, therefore, is that possibly it may be beneficial to try to treat patients with Fabry disease sooner to prevent this progression of the Gb3 accumulation.

There have been studies that have looked at the treatment of gastrointestinal symptoms in adults who have been treated with treatments for Fabry disease. Studies have shown that there is improvement in patients of their gastrointestinal symptoms. A little more than 50% of patients will have improvement of their gastrointestinal symptoms, with decrease in their diarrhea, and some patients have decrease in their abdominal pain.

However, it should also be noted that there are a significant amount of patients who will not have improvement in their symptoms. This leads one to think that possibly it may be beneficial to actually treat patients much earlier in their disease process, even prior to their symptom presentation. Pushing for more of a treatment process in very young patients as to prevent the manifestation of their gastrointestinal symptoms.