

SPEAKER: As studies have shown, many patients have improvement of their gastrointestinal symptoms with treatment of Fabry disease. However, there are a significant number of patients, a little less than half of them, who continue to have significant gastrointestinal symptoms even while being treated for their Fabry disease. Therefore, this leads us to think about how we can treat the gastrointestinal symptoms themselves and target therapy directly to the symptoms.

Based on the symptoms, there are various options for further workup and further treatment of these symptoms. When we think about the gastrointestinal symptoms, we think about similar treatments that might work for irritable bowel syndrome, which we think is related to some form of neuropathy, that might also treat patients with their Fabry disease.

For patients with diarrhea, there are various studies that can look at the motility of the gut, including a smart pill study, which can look at the full gut transit, and also a Sitzmark study, which looks at the transit of the colon. Both of these could give us more information about the transit time throughout the gut and can help lead want to target specific therapy to those who may have rapid or slow transit.

Treatment for this may include medications that will slow the transit time of the colon, including anti-diarrheal medications. Other various options might be treatment of the abdominal pain, which is thought to be due to neuropathy. There are various neuromodulators that have been used by gastroenterologists to treat neuropathic abdominal pain. These are thought to work on the nerves in the GI tract to decrease some of the abdominal pain that the patient may be experiencing.

Finally, there is also thought for the upper gastrointestinal symptoms, such as symptoms related to reflux disease or slow movement of the stomach. There are specific medications that are pro-motility medications that could help the early satiety and nausea that patients are feeling if they do have some form of gastroparesis. Additionally, proton pump inhibitors or antihistamines could be used to treat the reflux disease.