

SPEAKER: There is a patient who presented with a new diagnosis of Fabry disease who had abdominal pain, and their most debilitating symptom was severe diarrhea, up to 10 to 12 times a day where they would have to run to the bathroom with urgency, and also reported periods of incontinence. They would carry an extra set of clothes in their car because they wouldn't know when they would have the sudden urge to use the restroom. They also had issues with eating because that would frequently trigger the diarrhea, and so would plan their schedule around when they would eat based on when they would be near a bathroom.

They were started on treatment for Fabry disease with some improvement in their symptoms-- decrease in abdominal pain. However, the diarrhea continued even after several years of treatment. We were following them, and several years later, the diarrhea continued to be a persistent problem. However, decreased in frequency, but continued to have significant urgency with intermittent periods of abdominal pain.

Therefore, we started her on amitriptyline, which is a tricyclic antidepressant, which is used frequently by gastroenterologists for patients with irritable bowel syndrome diarrhea type, as it's thought to both target abdominal discomfort, and also slows down colonic transit so that patients with diarrhea can have some improvement in their symptoms. We started at a low dose and titrated the patient up to 50 milligrams. And she did very well with this-- continued to have looser stools, but decreased in frequency of the diarrhea around three times a day, and much improvement in the abdominal pain. This showed us that there was some improvement from her treatment of the Fabry disease, but also there was improvement with targeted therapy for the gastrointestinal symptoms too.