

SPEAKER: Though, the very young patients, it's actually fairly rare to see gastrointestinal manifestations. However, it has been reported, and some patients do have those presentations. However, it's very unlikely that it will be diagnosed at that age. It's more for the practitioners and the school-aged patients and middle school patients, where you'll have Fabry patients show up who have this recurring abdominal pain or significant diarrhea.

The diarrhea is something that should trigger some practitioners to think further into what might be causing it, as diarrhea is not as common at this age in patients. Unfortunately, abdominal pain is a frequent symptom in many, many patients without a genetic basis to it. And so in these patients who are presenting with frequent abdominal pain, a clinician needs to ask about other associated symptoms and about the family history.

Additionally, there is no other specific finding related to weight. That's always a question. There was a study that came out that showed that there was a difference in BMI and patients who had Fabry disease, as compared to their counterparts who did not have separate disease. However, a more recent study showed that actually the weight and the BMI are exactly the same in Fabry patients and those who don't have Fabry disease. Additionally, studies have shown-- sorry.

Additionally, the quality of life has been looked at in younger children with Fabry disease and found that those with Fabry disease have worse quality of life. There's some sot-- there's some thought that these patients also have significant gastrointestinal symptoms at this age, and therefore the GI symptoms might be leading to morbidity in these patients.

As for the provider, when a patient with Fabry disease shows up in your office without a diagnosis of Fabry disease, the most important way of diagnosing them is to look beyond their acute presentation of gastrointestinal symptoms and ask about chronicity, and also ask about the associated symptoms they might be having with it.