

SPEAKER: One of the less recognized complications of Fabry disease is a lymphedema. It usually presents with swelling in the feet and ankles and can get progressively more severe with time. It is much more common in association with classical Fabry disease, and generally more common, as well, as more severe in men and women. I think that it is an underappreciated complication.

In my clinic, we have about 100 patients with Fabry disease. And at least 15 to 20 of them have significant lymphedema that has required therapy. The lymphedema is a primary manifestation of the storage. We think that it's due to endothelial storage in the lymphatic vessels, as well as some vascular insufficiency developing in the smaller blood vessels.

It, however, is generally not associated with renal failure or heart failure at the time of the initial presentation. So use of diuretics is generally not going to be helpful in treating the lymphedema. The most effective therapy that I am aware of is lymphatic massage and restrictive clothing or wraps that would be used for other causes of primary lymphedema.