

SPEAKER: As Fabry Disease is a disease that develops with time and is it really has a very high mortality, we as a dermatologist have this very important role of early detection of Fabry Disease. Because we can change the life of this patient with the treatment. And that is very important. If we are able to diagnose them before they have-- before patients develop and stage disease or a renal disease or cardiovascular disease, we might really make a difference.

Then unfortunately, from a dermatologic point of view, we've seen that patients-- even with patients who will be able to start treatment very early, cutaneous manifestations don't change with time. And even patients that are stable on their disease, they might even carry on developing new angiokeratomas. That seems a little bit-- that's really a mystery for us. That's a little bit tricky. We don't know why that's it.

What we do know for example, is that cutaneous manifestations not always correspond with other organs manifestation. We have patients with lots of ventricular Thomas with a very broad diseased skin, with lesions everywhere. And not only in the typical distribution, but are very big and very hypercritical lesions that have nearly no systemic symptoms or very few systemic symptoms. But on the other hand, we've seen patients with very severe renal disease, cardiovascular disease, with very severe ophthalmologic disease. Or even with any other kind of manifestations that have very few cutaneous lesions. So cutaneous manifestations are not a very good biomarker of Fabry Disease. And as I've said, they don't respond very well to any of the two available treatments.

They carry on appearing on time. So it's not a good way to follow patients. So that's like saying these patients are doing well because they don't develop any new cutaneous lesions. Or this patient's not doing well because they still develop it. Because for the time being, cutaneous manifestations are not very well responding to the available treatments.

So once the patient's stable of the systemic disease, they sometimes require some kind of treatment. Of course at the beginning and with that diagnosis, skin lesions tend to be the less important. Patients don't care about them. They just want to be-- well, from the other aspects. But once they are stabilized, we can perform quite the different destructive methods. We need to use lasers, or we need to use other kinds of therapies such as kryotherapy, or light therapy for the destruction of these lesions.