SPEAKER: Angiokeratomas are very typical lesions. They are these red papules, these vascular papules, with a hyperkeratotic surface. In Fabry Disease, at the beginning, when patients start developing the lesions, they might not have this verruca surface. They might just look like cherry angiomas. But they tend to have this quite typical distribution along the bathing trunk area. Sometimes, and especially at the beginning, they might not show the typical distribution. And as lesions go appearing along time, you might just see a little or quite few of them if you are seeing a young patient.

So we as dermatologists are really at a position of being able to perform a biopsy. It's very easy to do a skin biopsy. Of course, a renal biopsy is very hard. A skin biopsy is very easy. And sometimes, in the histopathology, you might see-- the diagnosis might be angiokeratoma. And then you have to think about all the possibilities of all the diseases that might appear with angiokeratoma.

But sometimes-- and this is the tricky part-- lesions, especially when patients are very young, might not have this crusted area on the surface, this crusted keratosis from the angiokeratomas, and they might just show as telangiectasias. Then, if you don't have the histopathologic diagnosis after that, you might need to really rely on your clinical suspicion and ask about other symptoms, especially when patients are very young.

One very important thing to ask for is family history. If patients-- of course, you first have to see if the patient is a boy or the patient is girl, because it all depends on that. If you're thinking that the carrier or the parent who might have transmitted the disease is the mother or the father, but if you have a boy, then the only possibility is that he might have got it from the mother, so you should ask the mother about certain questions.

But probably, mothers don't have either the full-blown disease, because female patients might show a severe disease or a very mild disease. So you should ask some questions to the mother and, specifically, ask about family history of this typical Fabry Disease, the renal insufficiency, the cardiac manifestations, the neurologic manifestations, and all those manifestations. But not do not expect those manifestations to appear in a young kid.

In this young kid, you should ask about other manifestations, more subtle, those first manifestations that appear, that are the gastrointestinal manifestations, the neurologic, the small fiber manifestations, the acroparesthesias, and the fever of unknown origin. And sometimes, it's very helpful to send the patient to the ophthalmologist to see if they have any kind of ophthalmologic manifestations.

Again, the ophthalmologist should be someone who really knows about Fabry Disease, because cutaneous lesions might appear, might not be diagnosed in someone who sees them and does not know that they belong to Fabry Disease. The same thing might happen with the eye. So if you rely on someone, you should send them, the patient, to these specialized ophthalmologists. That might be very helpful in a young age, too.