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What shall we do when we, as dermatologists, suspect a patient might have Fabry's Disease? It all depends, of course, if it's a female patient or if it's a male patient. And if they have a family history or if they already have a diagnosis of a family member that has Fabry's Disease, or if they are the proband-- if they are the first patient.

What happens if we get to see a young adult patient, a male, with these angiokeratomas along the bathing suit area? And the first thing we should do is ask about other symptoms, about a history, a personal history of acroparesthesia, of gastrointestinal disease, and if they've ever been told that they have something atypical in their eyes. Because sometimes, as I've said, they are not recognized, and they don't have the specific name, but patients have been told they have something in their eyes, for example.

So if I have that patient, and he's a young adult, maybe he does not have, yet, developed any other symptoms. So then we should test for Fabry's Disease. Here in Argentina, we have the possibility of doing a dry drop, and after that, to confirm it in leukocytes, the activity, enzyme activity in leukocytes. And after that, we get the genetic testing.

Now, if it's a female patient, we know that female patients might not show the enzymatic alteration in blood. So we directly-- if we suspect of a Fabry's Disease, we should directly go to the genetic testing. And as I've said, then, if we see an older patient-- a little bit older. Not a very old patient, but someone who's-- a male who's around 30 or 40, then we, then, yes, suspect they should have other typical symptoms of Fabry's Disease or other typical signs of Fabry's Disease like renal disease, cardiovascular disease, or even stroke, cryptogenic stroke.

But maybe we should not focus ourselves, at that age, on, for example, acroparasthesias or this a small fiber pain, because sometimes, as time goes by, this pain tends to disappear because the nerves get so badly damaged, they don't even get to feel the pain. So if patients are a little bit older, then we don't ask, specifically, about acroparesthesias.

And what I do, because I work very near from an ophthalmologist who is specialized in Fabry's Disease is, once I've already got the blood drawn, in the same day, I send the patient to the ophthalmologist, and that's of lots of help. If he finds something else, then we have two cardinal signs of Fabry's Disease. And that, if we don't find the alterations in the blood or, even, in the genetic testing, then we insist. Because we sometimes know-- it's sometimes very easy, but sometimes it's hard to diagnose, even genetically, the disease.