

DR. PAULA

Angiokeratomas are the hallmark of Fabry disease, of the cutaneous manifestations of Fabry disease. These lesions, they are found, typically in Fabry patients. And they look like very tiny papules with a verrucous surface. Sometimes they look more like telangiectasias, especially in the first states of the disease. And then as time goes by, they tend to develop this hyperkeratotic surface. But they do look like-- I mean, for dermatologists, they are quite typical, but maybe for the other specialties, lesions look like red papules with a crust on top.

LUNA:

But these lesions that don't resolve spontaneously, that tend to appear over time. And although, most of Fabry patients-- especially male patients-- although, most of them have some kind of lesions, not everyone is able to recognize them. But we have to take into account that this is the hallmark of Fabry disease and we have to really, really look-- we have to make a very broad exam, dermatologic examination of the patients. And we are kind of always going to find them.

Even in females that we sometimes think they don't have manifestations or they have subtle manifestations, we have to look for these lesions. They are typically-- in female patients, they might appear, also in the genital area as in males, but they are more subtle. And umbilicus, the navel, is always a place to go to look for this kind of lesions.

As I've said, although the genetic alteration is present since birth, lesions tend to appear with time, but angiokeratomas are one of the first manifestations. But again, at the beginning, or when patients are very small, they might not be easily recognized. And they might not be present in the typical bathing trunk areas. Bathing trunk, meaning from the umbilicus to the upper thighs as they appear in full in the full blown disease.

And as I've said, they appear in most of the patients, especially in the classic Fabry disease. And it is interesting, too, if we realized that Fabry actually was a dermatologist who described the disease because he started describing it from the cutaneous manifestation. So that is very, very important. And angiokeratomas are very visual. If you see your patient, if you have another specialty, and you don't always see your patient fully naked, but at least ask the patient if they have this kind of lesions.