

SPEAKER: Angiokeratomas, with a typical distribution, usually described in later adolescence or in young adults, especially in males. They tend to appear a little bit earlier, but they are harder to diagnose because most of the times, they hadn't developed this hyperkeratotic lesion on top. So this red papules with the hyperkeratosis tends to appear around late adolescence or a young adult.

But lesions might be present since very early. With diagnosed patients, five or six years old, with subtle lesions, more telangiectatic like than typical angiokeratomas. But patients tend to have at this early age, other manifestations. Not the full blown disease. We're not expecting patients to have severe renal disease or cardiovascular disease, but these patients tend to have more subtle symptoms that we should ask about.

For example, gastrointestinal disease or acroparesthesias. If we ask a patient about, specifically acroparesthesias, they're not going to understand us, but we should ask if they have pain in their hands, in their feet. If they feel like there's needles and pins or if they feel this fire in their hands and feet. And if this patient, they might also show or have fever of unknown origin because the same deposition happens in the sweat glands that might not allow the patient to sweat correctly and that causes sometimes fevers. So younger patients might show lesions, subtle angiokeratomas but also gastrointestinal disease, fever of unknown origin, and this neurologic pain.