GENETICS AND GENODERMATOSES

EXUBERANT CASE OF PSEUDOXANTHOMA ELASTICUM

Monallisa Lima Andrade [(1)] - Paula Santana Alves Carneiro [(1)] - Duanne Silva Freitas [(1)] - Felipe Batista Santos [(1)] - Bruno De Santana Silva [(1)] - Fedro Menezes Portugal [(1)] - Pedro Dantas Oliveira [(1)]

Federal University Of Sergipe, Department Of Medicine, Aracaju, Brazil [(1)]

Background: Pseudoxanthoma elasticum (PXE) is an autosomal recessive disease of the connective tissue, characterized by mutations in the ABCC6 gene complex. The lack of functional ABCC6 protein leads to ectopic mineralization that is most apparent in the elastic tissues of the skin, blood vessels and eyes. Clinical manifestations are generally absent at birth and the skin lesions start to develop in the first or second decade. The first clinical sign of PXE is almost always small yellow papules on the nape and sides of the neck and in flexural areas. The papules coalesce, and the skin becomes loose and wrinkled. Histologically, elastorrhexis is the most important histological feature of PXE, stained with Verhoeff-Van Gieson. The mid-dermal elastic fibers are short, fragmented, clumped and calcified. PXE patients may have premature atherosclerosis, caused by the mineralization and fragmentation of the elastic fibers of the medium-sized arteries and the aorta. Furthermore, occurs dystrophic calcification of Bruch's membrane, which mays trigger choroidal neovascularization and blindness in late-stage disease.

Observation: A 57-year-old female patient reports on the appearance of asymptomatic lesions in the lateral regions of the neck and underarms since childhood. It presents hypertension, peripheral vascular disease and reduction of visual acuity. At the examination, yellowish coalescent papules were present in the lateral regions of the neck and underarms, with changes in the elasticity of the skin. A skin biopsy revealed a superficial and deep dermis with basophilic elastic fibers, short, fragmented and clumped, stained by Verhoeff-Van Gieson, compatible with PXE.

Key message: There is no cure for PXE and is associated with a risk of decreased quality of life, peripheral vascular compromise and blindness. Patients should be monitored on a regular basis and more studies are needed for disease-modifying treatments.