Opis choroby *

Definicja

A rare, genetic, dermis elastic tissue disorder characterized by yellowish skin papules (resembling pseudoxanthoma elasticum) located on the neck, chest and/or flexural areas associated with loose, redundant, sagging skin on trunk and upper limbs, and retinitis pigmentosa, in the absence of clotting abnormalities. Patients present reduced night and peripheral vision, as well as optic nerve pallor, retinal pigment epithelium loss, attenuated retinal vessels and/or black pigment intra-retinal clumps.

Dane

Klasyfikacja
Choroba

Synonimy
PXE-like syndrome with retinitis pigmentosa
Zespół podobny do PXE ze zwyrodnieniem barwnikowym siatkówki

Kod ORPHA
436274

Kod OMIM
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Kod ICD10
Q82.8

Kod ICD11
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*Źródło

orphanet