

Opis choroby *

Definicja

A rare acquired dermis elastic tissue disorder characterized by clinical and histopathologic evidence of pseudoxanthoma elasticum in the absence of a family history or specific mutation. Patients present with predominantly cutaneous manifestations consisting of yellowish papules which coalesce into large plaques and are most commonly localized on the neck, axillae, groin, and flexural surfaces. Skin biopsy shows accumulation of clumped, calcified elastic fibers in the mid-dermis. Reported underlying factors include previous liver transplantation, exposure to penicillamine, or concomitant beta-thalassemia.

Dane

Klasyfikacja

Choroba

Synonimy

Acquired Gronblad-Strandberg-Touraine syndrome
Nabyte PXE
Nabyty zespół Gronblada, Strandberga i Touraine'a
Acquired PXE

Kod ORPHA

228247

Kod OMIM

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Kod ICD10

L98.8

Kod ICD11

EE41.Y

*Źródło

orphanet