Opis choroby *

Definicja

A rare, genetic, isolated diffuse palmoplantar keratoderma characterized by diffuse, mild to thick, finely demarcated hyperkeratosis of palms and soles. Additional clinical findings include knuckle pad-like keratoses on fingers, hyperkeratosis of umbilicus and areolae, diffuse dry skin, hyperhidrosis, hangnails and frequent fungal infections. Histological examination of lesions reveals orthokeratotic hyperkeratosis, acanthosis, hypergranulosis, and mild lymphocyte infiltrations in the upper dermis with no evidence of epidermolysis.

Dane

Klasyfikacja
Choroba

Synonimy
KRT1-related diffuse NEPPK
KRT1-related diffuse NEPPK

Kod ORPHA
530838

Kod OMIM
600962

Kod ICD10
Q82.8

Kod ICD11
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*Źródło

orphanet