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 Synonimy

Choroba KRT1-related diffuse NEPPK

KRT1-related diffuse NEPPK

 Kod ORPHA
 Kod OMIM
 Kod ICD10

 530838
 600962
 Q82.8

Kod ICD11

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*Źródło

orphanet