

Opis choroby *

Definicja

A rare, genetic punctate palmoplantar keratoderma disease characterized by discrete, focal, punctate keratoderma on the palms and soles and/or slowly progressive spastic paralysis, predominantly affecting the lower limbs. Lesional histology reveals pronounced orthokeratosis, acanthosis, papillomatosis, and regular undulation to the surface keratin. There have been no further descriptions in the literature since 1983.

Dane

Klasyfikacja

Choroba

Synonimy

Palmoplantar hyperkeratosis-spastic paralysis syndrome
Hiperkeratoza dłoni i stóp-zespół porażenia spastycznego
Zespół Powella, Venencie i Gordona
Powell-Venencie-Gordon syndrome

Kod ORPHA

2201

Kod OMIM

148360

Kod ICD10

Q82.8

Kod ICD11

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*Źródło

orphanet