

Opis choroby *

Definicja

Palmoplantar keratoderma-deafness syndrome is a keratinization disorder characterized by focal or diffuse palmoplantar keratoderma. A patchy distribution is observed with accentuation on the thenars, hypothenars and the arches of the feet. The disease becomes apparent in infancy and is associated with sensorineural hearing loss that shows a variable age of onset. Due to genetic and clinical similarities, it has been proposed that palmoplantar keratoderma-deafness syndrome, knuckle pads-leukonychia-sensorineural deafness-palmoplantar hyperkeratosis syndrome and keratoderma hereditarium mutilans may represent variants of one broad disorder of syndromic deafness with heterogeneous phenotype. The disease is transmitted in an autosomal dominant manner with incomplete penetrance.

Dane

Klasyfikacja

Choroba

Synonimy

PPK-deafness syndrome
Zespół hiperkeratoza dłoni i stóp - głuchota
Zespół hiperkeratoza dłoni i stóp - utrata słuchu
Zespół PPK-głuchota
Zespół rogowiec dłoni i stóp - utrata słuchu
Palmoplantar hyperkeratosis-deafness syndrome
Palmoplantar hyperkeratosis-hearing loss syndrome
Palmoplantar keratoderma-hearing loss syndrome

Kod ORPHA

2202

Kod OMIM

148350

Kod ICD10

Q82.8

Kod ICD11

EC20.30

*Źródło

orphanet

