

Opis choroby *

Definicja

A rare, syndromic genetic deafness disease characterized by symmetric or asymmetric knuckle pads (typically located on the distal and interphalangeal joints), leukonychia, diffuse palmoplantar keratoderma, and congenital, mild to moderate sensorineural deafness.

Dane

Klasyfikacja

Choroba

Synonimy

Bart-Pumphrey syndrome

Zespół Barta i Pumphrey'a

Zespół knuckle pads-leukonychia-głuchota

czuciowo-nerwowa-hiperkeratoza dłoni i stóp

Knuckle pads-leukonychia-sensorineural

deafness-palmoplantar keratoderma syndrome

Knuckle pads-leukonychia-sensorineural hearing

loss-palmoplantar hyperkeratosis syndrome

Knuckle pads-leukonychia-sensorineural hearing

loss-palmoplantar keratoderma syndrome

Kod ORPHA

2698

Kod OMIM

149200

Kod ICD10

Q82.8

Kod ICD11

LD2H.Y

*Źródło

orphanet