

## 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

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#### \*Źródło

orphanet