

Opis choroby *

Definicja

A rare genetic syndrome with limb duplication, polydactyly, syndactyly, and/or hyperphalangy characterized by duplication anomalies such as triphalangeal thumbs, phalangeal duplication of other digits, and polydactyly, associated with highly variable combinations of ectrodactyly, brachydactyly, and syndactyly of hands and/or feet. Severe nail dysplasia or absence of nails is also observed.

Dane

Klasyfikacja

Zespół wad wrodzonych
Synonimy
Carnevale-Hernández-del Castillo syndrome
Zespół Carnevale'a, Hernandeza i del Castillo

Kod ORPHA

2947

Kod OMIM

190680

Kod ICD10

Q74.8

Kod ICD11

LD26.2

*Źródło

orphanet