

Opis choroby *

Definicja

A rare congenital limb malformation syndrome characterized by hypoplasia or aplasia of the terminal parts of fingers 2 to 5, with complete absence of the fingernails. The thumbs are always intact but frequently show flattening, splitting or duplication of the distal phalanges. Digits on the radial side of the hand are less severely affected than those on the ulnar side. The feet are similarly affected but less severely. Soft tissue syndactyly, symphalangism, carpal and/or tarsal fusions and shortening of metacarpals and/or metatarsals may be present.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

93383

Kod OMIM

113000

Kod ICD10

Q73.8

Kod ICD11

LD26.1

*Źródło

orphanet