Opis choroby *

Definicja

A rare ectodermal dysplasia syndrome characterized by neonatal teeth, trichodystrophy (with straw-like, discolored and fragile hair), onychodystrophy, and malformation of the hands and feet consisting of simian-like hands with transverse palmar creases and prominent interdigital folds, brachydactyly, and marked shortness of the first metacarpal and metatarsal bones with hypoplasia of the distal phalanges. There have been no further descriptions in the literature since 1997.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
OTUDP syndrome
Zespół OTUDP
Zespół zębowo-włosowo-paznokciowo-palcowo-dłoniowy, typ Mendoza i Valiente
Odonto-tricho-ungual-digito-palmar syndrome, Mendoza-Valiente type

Kod ORPHA
69082

Kod OMIM
601957

Kod ICD10
Q82.4

Kod ICD11
LD27.0Y

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*Źródło

orphanet