Opis choroby *

Definicja

A rare ectodermal dysplasia syndrome characterized by the association of hypocalcified and hypoplastic tooth enamel, distal finger and toenail onycholysis with subungueal hyperkeratosis, and functional hypohidrosis. Additional manifestations include seborrheic scalp dermatitis and rough, dry skin. Lacrymal punctae may be occasionally absent. There have been no further descriptions in the literature since 1975.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Ameloonychohypohidrotic ectodermal dysplasia
Ameloonychohypohidrotic syndrome

Kod ORPHA
1028

Kod OMIM
104570

Kod ICD10
Q82.4

Kod ICD11
LD27.0Y

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*Źródło

orphanet