

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

Synonimy

Zespół wad wrodzonych Ameloonychohypohidrotic ectodermal dysplasia
Ameloonychohypohidrotic syndrome

Kod ORPHA

1028

Kod OMIM

104570

Kod ICD10

Q82.4

Kod ICD11

LD27.0Y

*Źródło

orphanet