Opis choroby *

Definicja

A rare, genetic, ectodermal dysplasia syndrome characterized by the association of hypohidrotic ectodermal dysplasia (manifesting with the triad of hypohidrosis, anodontia/hypodontia and hypotrichosis) with primary hypothyroidism and respiratory tract ciliary dyskinesia. Patients frequently present urticaria pigmentosa-like skin pigmentation, increased mast cells and melanin depositions in the dermis and severe, recurrent chest infections. There have been no further descriptions in the literature since 1986.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
ANOTHER syndrome
Zespół HEDH
Zespół ANOTHER
HEDH syndrome

Kod ORPHA
1882

Kod OMIM
225050

Kod ICD10
Q82.4

Kod ICD11
LD27.02

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*Źródło

orphanet