## 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