

## 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 ANOTHER syndrome

#### Synonimy

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