

## Opis choroby \*

### Definicja

A rare ectodermal dysplasia syndrome characterized by anonychia congenita totalis or rudimentary nails, macular hyper- and/or hypopigmentation (particularly affecting groins, axillae and breasts), coarse scalp hair (that becomes markedly thinned in early adult life), dry palmoplantar skin with distorted epidermal ridges and sore, cracked soles, and hypohidrosis. There have been no further descriptions in the literature since 1975.

### Dane

### Klasyfikacja

Zespół wad wrodzonych

#### Kod ORPHA

69125

#### Kod OMIM

106750

#### Kod ICD10

Q84.3

#### Kod ICD11

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### \*Źródło

orphanet