

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