

Opis choroby *

Definicja

Dermotrichic syndrome is a rare, genetic, ectodermal dysplasia syndrome characterized by skin, hair and nail anomalies (i.e. generalized ichthyosis, congenital alopecia universalis, dystrophic, convex nails), associated with hypohidrosis without hyperthermia, intellectual disability, seizures, and skeletal (e.g. proportionate short stature, platyspondyly) and intestinal (e.g. congenital aganglionic megacolon) anomalies. Facial dysmorphism includes frontal bossing, blepharophimosis, large ears, low nasal bridge and small nose. There have been no further descriptions in the literature since 1992.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA	Kod OMIM	Kod ICD10
99688	-	Q82.4
Kod ICD11		
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*Źródło

orphanet