

Opis choroby *

Definicja

A rare ectodermal dysplasia syndrome characterized by congenital onychodystrophy (particularly of the distal nail) and severe hypotrichosis with alopecia involving the eyebrows, eyelashes and body hair. Scalp, beard, pubic and axillary hair is brittle and shows a twisting pattern on electron microscopy. There have been no further descriptions in the literature since 1991.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA	Kod OMIM	Kod ICD10
2890	-	Q82.4

Kod ICD11

LD27.0Y

*Źródło

orphanet