

## 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**  
2890

**Kod OMIM**  
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**Kod ICD10**  
Q82.4

**Kod ICD11**  
LD27.0Y

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### \*Źródło

orphanet