Opis choroby *

Definicja

A rare ectodermal dysplasia syndrome characterized by bilateral retinitis pigmentosa, trichodysplasia (generalized hypotrichosis, structural changes), dental anomalies, onychodysplasia, and dry and scaly skin. There have been no further descriptions in the literature since 1988.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Cecato de Lima-Pinheiro syndrome
Zespół Cecato de Lima i Pinheiro

Kod ORPHA
2718

Kod OMIM
257960

Kod ICD10
Q82.4

Kod ICD11
LD27.0Y

---

*Źródło

orphanet