

Opis choroby *

Definicja

Trichodermodyplasia-dental alterations syndrome is a rare, genetic ectodermal dysplasia syndrome characterized by sparse, thin, brittle scalp hair, as well as sparse eyebrows, eyelashes, axillary and pubic hair, delayed eruption of deciduous teeth and hypodontia of both dentitions. Mild palmoplantar keratosis, café-au-lait spots on back, mild dystrophy of nails, and tibial deflection of toes are also associated. There have been no further descriptions in the literature since 1986.

Dane

Klasyfikacja

Synonimy

Zespół wad wrodzonych Pinheiro-Freire Maia-Miranda syndrome
Zespół Pinheiro, Freire Maia i Miranda

Kod ORPHA
3353

Kod OMIM
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Kod ICD10
Q82.4

Kod ICD11
LD27.0Y

*Źródło

orphanet