Opis choroby *

Definicja

A rare ectodermal dysplasia syndrome characterized by hypotrichosis of scalp and eyebrows, finger syndactyly, intellectual disability and early eruption of teeth. Facial dysmorphism (i.e. round face with prominent forehead, cheeks and ears, and upward-slanting palpebral fissures), hypoplasia of median and distal phalanges, and kyphosis are additionally observed features. There have been no further descriptions in the literature since 1996.

Dane

Klasyfikacja
Choroba

Synonimy
Lopes-Marques de Faria syndrome
Zespół Lopesa i Marques de Faria

Kod ORPHA
2266

Kod OMIM
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Kod ICD10
Q82.4

Kod ICD11
-

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*Źródło

orphanet