

Opis choroby *

Definicja

Ichthyosis-oral and digital anomalies syndrome is characterised by ichthyosis, unusual facies (small mouth with a thin upper lip and lower lip with a midline groove) and digital anomalies (tapered fingers with a lack of distal flexion creases and wide spacing between the second and third fingers). It has been described in two sibs born to first cousin parents. Transmission appears to be autosomal recessive.

Dane

Klasyfikacja

Zespół wad wrodzonych Clayton Smith-Donnai syndrome
Zespół Clayton-Smith i Donnai

Synonimy

Kod ORPHA

2272

Kod OMIM

258840

Kod ICD10

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Kod ICD11

LD27.2

*Źródło

orphanet