

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

Kod ORPHA	Kod OMIM	Kod ICD10
2272	258840	-

Kod ICD11
LD27.2

*Źródło

orphanet