

Opis choroby *

Definicja

Hirschsprung disease-type D brachydactyly syndrome is characterized by Hirschsprung disease and absence or hypoplasia of the nails and distal phalanges of the thumbs and great toes (type D brachydactyly). It has been described in four males from one family (two brothers and two maternal uncles). Transmission appears to be X-linked recessive but autosomal dominant inheritance with incomplete penetrance in females can not be ruled out.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

2150

Kod OMIM

306980

Kod ICD10

Q43.1

Kod ICD11

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*Źródło

orphanet