Choroba Hirschsprunga - brachydaktylia typu D Kod Orpha: 2150 Kod OMIM: 306980

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

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.