

## 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