

## Opis choroby \*

### Definicja

Hirschsprung disease-nail hypoplasia-dysmorphism syndrome is a fatal malformative disorder that is characterized by Hirschsprung disease, hypoplastic nails, distal limb hypoplasia and minor craniofacial dysmorphic features (flat facies, upward slanting palpebral fissures, narrow philtrum, narrow, high arched palate, micrognathia, low set ears with abnormal helices). Hydronephrosis has also been reported. There have been no further descriptions in the literature since 1988.

### Dane

#### Klasyfikacja

Zespół wad wrodzonych Al Gazali-Donnai-Muller syndrome  
Zespół Al Gazali, Donnai i Mullera

#### Synonimy

#### Kod ORPHA

2153

#### Kod OMIM

235760

#### Kod ICD10

Q43.1

#### Kod ICD11

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#### \*Źródło

orphanet