

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

Kod ORPHA	Kod OMIM	Kod ICD10
2153	235760	Q43.1

Kod ICD11

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*Źródło

orphanet