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Hirschsprung disease-nail hypoplasia-dysmorphism syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base.

Hirschsprung disease-nail hypoplasia-dysmorphism syndrome. ORPHA:2153

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.