Hirschsprung disease-deafness-polydactyly syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hirschsprung disease-deafness-polydactyly syndrome. ORPHA:2155

Hirschsprung disease-deafness-polydactyly syndrome is an extremely rare malformative association, described in only two siblings to date, characterized by Hirschsprung disease (defined by the presence of an aganglionic segment of variable extent in the terminal part of the colon that leads to symptoms of intestinal obstruction, including constipation and abdominal distension), polydactyly of hands and/or feet, unilateral renal agenesis, hypertelorism and congenital deafness. There have been no further descriptions in the literature since 1988.