Opis choroby *

Definicja

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.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Hirschsprung disease-hearing loss-polydactyly syndrome
Zespół Santosa, Mateusa i Leala
Santos-Mateus-Leal syndrome

Kod ORPHA
2155

Kod OMIM
235740

Kod ICD10
Q43.1

Kod ICD11
LD2H.Y

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*Źródło

orphanet