Opis choroby *

Definicja

A rare, genetic, developmental defect during embryogenesis syndrome characterized by total or partial colonic aganglionosis associated with peripheral, usually multifocal, neuroblastic tumors (ganglioneuroblastoma, neuroblastoma, ganglioneuroma). Congenital central hypoventilation syndrome, with variable severity of respiratory compromise, cardiovascular and ophthalmologic symptoms, consistent with autonomic nervous system dysfunction, is occasionally associated.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
2151

Kod OMIM
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Kod ICD10
Q43.1

Kod ICD11
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*Źródło

orphanet