Hirschsprung disease-ganglioneuroblastoma syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hirschsprung disease-ganglioneuroblastoma syndrome. ORPHA:2151

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.