Goldberg-Shprintzen megacolon syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Goldberg-Shprintzen megacolon syndrome. ORPHA:66629

A rare multiple congenital anomalies/dysmorphic syndrome characterized by Hirschsprung disease, facial dysmorphism (sloping forehead, high arched eyebrows, long eyelashes, telecanthus/hypertelorism, ptosis, prominent ears, thick earlobes, prominent nasal bridge, thick philtrum, everted lower lip vermillion and pointed chin), global developmental delay, intellectual disability and variable cerebral abnormalities (focal or generalized polymicrogyria, or hypoplastic corpus callosum).