Cortical blindness-intellectual disability-polydactyly syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Cortical blindness-intellectual disability-polydactyly syndrome. ORPHA:1389

A rare, genetic, multiple congenital anomalies/dysmorphic syndrome characterized by congenital, total, cortical blindness, intellectual disability, postaxial polydactyly of the hands and feet, pre- and postnatal growth delay, psychomotor developmental retardation, and mild facial dysmorphism (incl. prominent forehead, short nose, long philtrum, high-arched palate, and microretrognathia). Recurrent respiratory and intestinal infections, as well as moderate hypertonia and hyperreflexia, are also associated. There have been no further descriptions in the literature since 1985.