Pontine tegmental cap dysplasia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Pontine tegmental cap dysplasia. ORPHA:269229

Pontine tegmental cap dysplasia is a rare, central nervous system malformation characterized by specific pattern of congenital anomalies affecting the pons, medulla, and cerebellum. Clinical manifestations of multiple cranial nerves deficits, pyramidal and cerebellar signs include neonatal hypotonia, ataxia, sensorineural deafness, reduced vision, language and speech disorders, feeding and swallowing difficulties, facial paralysis and intellectual disability. Various cardiac, gastrointestinal, genitourinary and skeletal defects have been sometimes reported.