Port-wine nevi-mega cisterna magna-hydrocephalus syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Port-wine nevi-mega cisterna magna-hydrocephalus syndrome. ORPHA:2703

A rare developmental defect during embryogenesis syndrome characterized by a glabellar capillary malformation, congenital communicating hydrocephalus, and posterior fossa brain abnormalities, including Dandy-Walker malformation, cerebellar vermis agenesis, and mega cisterna magna. Seizures are occasionally associated. There have been no further descriptions in the literature since 1979.