Pontocerebellar hypoplasia type 6

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Pontocerebellar hypoplasia type 6. ORPHA:166073

Pontocerebellar hypoplasia type 6 (PCH6) is a rare form of pontocerebellar hypoplasia (see this term) characterized clinically at birth by hypotonia, clonus, epilepsy impaired swallowing and from infancy by progressive microencephaly, spasticity and lactic acidosis.