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Pontocerebellar hypoplasia type 4

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base.

Pontocerebellar hypoplasia type 4. ORPHA:166063

Pontocerebellar hypoplasia type 4 (PCH4) is a very rare form of PCH (see this term), characterized by prenatal onset of polyhydramnios and contractures followed by hypertonia, severe clonus, primary hypoventilation leading to an early postnatal death.