Pontocerebellar hypoplasia type 5

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Pontocerebellar hypoplasia type 5. ORPHA:166068

Pontocerebellar hypoplasia type 5 (PCH5) is a very rare severe form of PCH (see this terme) with prenatal onset and characterized by fetal onset of clonus or seizures-like activity persisting in infancy and microencephaly leading to early postnatal death. There is significant overlap both in phenotype and in genotype between pontocerebellar hypoplasia types 4 and 5.