Non-syndromic pontocerebellar hypoplasia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Non-syndromic pontocerebellar hypoplasia. ORPHA:98523

Nonsyndromic pontocerebellar hypoplasias (PCH) are a rare heterogeneous group of diseases characterized by hypoplasia and atrophy and/or early neurodegeneration of the cerebellum and pons. Eight subtypes named type 1-8 have been described (see these terms), generally inherited in an autosomal recessive pattern.