Pontocerebellar hypoplasia type 2

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Pontocerebellar hypoplasia type 2. ORPHA:2524

Pontocerebellar hypoplasia type 2 (PCH2) is the most common subtype of pontocerebellar hypoplasia (see this term) characterized by neonatal onset and a lack of voluntary motor development and later progressive microencephaly, generalized clonus, development of chorea and spasticity. The majority of patients will not reach puberty.