

[Open Peer Review on Qeios](#)

# 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.