Congenital neuronal ceroid lipofuscinosis

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Congenital neuronal ceroid lipofuscinosis. ORPHA:168486

Congenital neuronal ceroid lipofuscinosis (CNCL) is a severe form of neuronal ceroid lipofuscinosis (NCL; see this term) with onset at birth characterized by primary microcephaly, neonatal epilepsy, and death in early infancy.