Niemann-Pick disease type C

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Niemann-Pick disease type C. ORPHA:646

Niemann-Pick disease type C (NP-C) is a lysosomal lipid storage disease (see this term) characterized by variable clinical signs, depending on the age of onset, such as prolonged unexplained neonatal jaundice or cholestasis, isolated unexplained splenomegaly, and progressive, often severe neurological symptoms such as cognitive decline, cerebellar ataxia, vertical supranuclear gaze palsy (VSPG), dysarthria, dysphagia, dystonia, seizures, gelastic cataplexy, and psychiatric disorders.