Niemann-Pick disease type A

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Niemann-Pick disease type A. ORPHA:77292

Niemann-Pick disease type A is a very severe subtype of Niemann-Pick disease, an autosomal recessive lysosomal disease, and is characterized clinically by onset in infancy or early childhood with failure to thrive, hepatosplenomegaly, and rapidly progressive neurodegenerative disorders.