



Pediatric Neurology Part III: Chapter 176. Niemann-Pick diseases (Handbook of Clinical Neurology)

Marie T. Vanier

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The Niemann–Pick disease group is now divided into two distinct entities: (1) acid sphingomyelinase-deficient Niemann–Pick disease (ASM-deficient NPD) resulting from mutations in the *SMPD1* gene and encompassing type A and type B as well as intermediate forms; (2) Niemann–Pick disease type C (NP-C) including also type D, resulting from mutations in either the *NPC1* or the *NPC2* gene. Both Niemann–Pick diseases have an autosomal recessive inheritance and are lysosomal lipid storage disorders, with visceral (type B) or neurovisceral manifestations. The clinical knowledge is updated taking into account recent surveys in large cohort of patients, particularly for type B and type C. The diagnosis of NP-C is often delayed due to the wide spectrum of clinical phenotypes. Systemic manifestations, if present, always precede onset of neurological manifestations. Most common neurological signs are vertical supranuclear gaze palsy, cerebellar ataxia, dysarthria, dysphagia, and progressive dementia. Cataplexy, seizures, and dystonia are other common features of NP-C. For both ASM-deficient NPD and NP-C, strategies for laboratory diagnosis of patients and prenatal diagnosis are discussed. Recent progress towards enzyme replacement therapy in type B patients and management of the neurological disease in type C patients are finally highlighted.



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