

RISK PREDICTION FOR NIEMANN-PICK DISEASE

A retrospective chart review of 216 patients with Niemann-Pick disease type C (NP-C) was conducted in 5 centers in Europe including University of Amsterdam and 2 in Australia. Three patient types were selected: classic or variant filipin staining NP-C cases (n=71) including family members with NP-C, NP-C filipin-negative staining noncases (n=64), or controls with at least 1 characteristic symptom of NP-C (n=81). NP-C symptoms and signs were categorized into visceral, neurologic, or psychiatric domains. Logistic regression was performed on individual signs and symptoms within and across domains, and regression coefficients were used to develop prediction scores for NP-C.

The suspicion index tool has good discriminatory performance, and patients with a score >70 should be tested for NP-C. Strong predictors of NP-C are neonatal jaundice/cholestasis, splenomegaly, vertical supranuclear gaze palsy, cataplexy, and cognitive decline/dementia; also, symptoms occurring in multiple domains in individual patients, and parents/siblings or cousins with NP-C. (Wijburg FA, Sedel F, Pineda M, et al. Development of a suspicion index to aid diagnosis of Niemann-Pick disease type C. *Neurology* May 15;78(20):1560-1567). (Response and reprints: Dr Wijburg. E-mail: f.a.wijburg@amc.uva.nl).

COMMENT. NP-C is a rare inherited neurovisceral disease caused by mutations in the NPC1 (95%) or NPC2 gene (5% cases) that lead to accumulation of cholesterol and glycosphingolipids in the brain, liver and other tissues. Foamy cells are present in the bone marrow, spleen and liver, and sea-blue histiocytes in the bone marrow. Neurological manifestations include saccadic eye movement abnormalities or vertical supranuclear gaze palsy, cerebellar ataxia, dystonia, dysmetria, dysarthria and dysphagia, gelastic cataplexy, and seizures. Age at presentation is early-infantile, late infantile, juvenile or adolescent/adult. International guidelines for management of NP-C were published in 2009, updated 2011. Disease-specific therapy with miglustat is reevaluated. (Patterson MC et al. *Mol Genet Metab* 2012 May 7. Epub ahead of print).

Four varieties of NP disease are distinguished, ABC & D. Type A is the classic infantile neuronopathic form, presenting with failure to thrive, persistent neonatal jaundice, hepatomegaly, lymphadenopathy, and sometimes a retinal cherry red spot. It is more common in Ashkenazi Jewish families. Type D is found in Nova Scotian families.

AUTOIMMUNE AND DEMYELINATING DISORDERS

MYASTHENIA GRAVIS AND NEUROMYELITIS OPTICA ASSOCIATION

Investigators at the University of Oxford, UK and 8 other international neurology centers describe the clinical, serological, and temporal associations of myasthenia gravis (MG) and neuromyelitis optica spectrum disorder (NMOSD) in 16 patients. All had early onset acetylcholine receptor antibody [AChR-Ab]-mediated MG, the majority with mild generalized disease, and a high proportion achieved remission. The MG preceded