Vitamin E and Niemann–Pick Disease Type C

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Niemann–Pick disease type C (NPC) is a lysosomal storage disorder, an autosomal recessive disease with an estimated incidence of 1 : 150,000, caused by mutations in the NPC1 or NPC2 proteins. The disease manifests in ataxia, seizures, and early dementia, and is typically fatal. In addition to aberrant trafficking of cholesterol, the levels and cellular distribution pattern of α-tocopherol are abnormal in brains and livers of NPC mouse models. Currently, there is no cure for NPC and all established therapies are for relief of symptoms with limited efficacy. Clinical trials with Miglustat are in progress with favorable preliminary results. Moreover, the ‘trapped’ α-tocopherol is not available for cellular use, and thought to contribute to the increased oxidative stress observed in NPC models, and likely underlie the anatomic and functional decline of the cerebellum seen in NPC. The notion that defective vitamin E status is an important factor in NPC disease is supported by studies showing that vitamin E (δ-tocopherol) supplementation improves cognitive function in mouse models of NPC disease with reducing the accumulation of free cholesterols and reduction of the size of enlarged lysosomes.

Keywords: Vitamin E; NPC; Lysosomal storage disease

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