

Compositions and Methods for the Treatment of Diseases associated with Faulty Cholesterol Regulation

Background

Niemann-Pick disease is a group of lipid storage disorder that affects cholesterol metabolism, with types A, B and C (NP-A, NP-B, and NP-C) being the most common forms of the disease. NP-A and NP-B are autosomal recessive disorders caused by a deficiency of the enzyme sphingomyelinase. Patients with NP-A or NP-B are unable to metabolize sphingomyelin leading to an accumulation of this lipid within the cell. Patients with NP-A and NP-B exhibit drastically different clinical manifestations, with NP-A patients exhibiting severe neurological effects and patients with NP-B patients having no neurological problems. NP-C is an autosomal recessive disorder characterized by central nervous system and visceral symptoms, and premature death. Patients with NP-C have faulty cholesterol transport, which results in the accumulation of LDL-derived cholesterol within the lysosome and increase in endogenous cholesterol synthesis. Furthermore, the feedback cycle for the down regulation of cholesterol synthesis is defective leading to a further accumulation of cholesterol within the cell. The gene responsible for NP-C1 has been identified and mapped to chromosome 18q11 while the gene responsible for NP-C2 remains unknown. **The present invention identifies HE1 as the gene responsible for NP-C2 disease.**

Description of the Technology

Studies at UMDNJ laboratory have identified HE1, as the gene responsible for NP-C2. HE1 has been previously identified as a cholesterol binding protein isolated from epididymal fluid and is a ubiquitously expressed lysosomal protein. A defect in HE1 protein expression was identified as the cause of accumulation of LDL-derived cholesterol in NP-C2 patients. Furthermore, addition of HE1 protein to NP-C2 fibroblasts lead to the reversal of the cholesterol accumulation in these cells. This discovery provides a basis for the therapy and diagnosis of NP-C2 and other diseases linked to faulty cholesterol regulation such as Alzheimer's disease, diabetes, and cardiovascular disease.

Applications

- Therapy of diseases linked to faulty cholesterol regulation such as Alzheimer's disease, diabetes, and cardiovascular disease.
- Diagnosis of diseases linked to faulty cholesterol regulation such as Alzheimer's disease, diabetes, and cardiovascular disease.

Patent Status

- US patent application filed
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Licensing Opportunity

This technology is available for licensing exclusively.

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