Genes For Niemann-Pick Type C Disease

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Technology description

Summary

Niemann-Pick disease is a class of inherited lipid storage diseases. Niemann-Pick Type C disease is an autosomal recessive neurovisceral lipid storage disorder which leads to systemic and neurological abnormalities including ataxia, seizures, and loss of speech. Patients with the disease typically die as children. The biochemical hallmark of Niemann-Pick Type C cells is the abnormal accumulation of unesterified cholesterol in lysosomes, which results in the delayed homeostatic regulation of both uptake and esterification of low density lipoprotein (LDL) cholesterol. Niemann-Pick Type C is characterized by phenotypic variability. The disease appears at random in families that have no history of the disorder, making diagnosis problematic. This invention provides the human gene for Niemann-Pick Type C disease and the nucleic acid sequences corresponding to the human gene for Niemann-Pick Type C disease. Also provided is the mouse homolog of the human gene. The invention could lead to improved diagnosis and the design of therapies for the disease and improved means of detection of carriers of the gene. In addition, this invention may contribute to the understanding and development of treatments for atherosclerosis, a more common disorder associated with cholesterol buildup that involves the accumulation of fatty tissue inside arteries that blocks blood flow, leading to heart disease and stroke. The invention may also lead to additional discoveries concerning how cholesterol is processed in the body.

Institution

NIH - National Institutes of Health

联系我们

叶先生

电 话：021-65679356

手 机：13414935137

邮 箱：yeyingsheng@zf-ym.com