Oxysterol Analysis for the Diagnosis of Niemann-Pick C1 Disease

Summary / Zusammenfassung

Niemann-Pick type C1 (NPC1) disease is a type of lysosomal storage disorders (LSD) characterized by accumulation of cholesterol in the endolysosomes. This rare progressive neurodegenerative disorder mainly results from mutations in the NPC1 gene. The condition leads to impairment of motor and intellectual functions and mostly to death occurring usually in adolescence. Treatments for NPC1 disease, such as substrate reduction therapy, are becoming available but early diagnostic and start of therapy is essential for the life expectancy and quality of NPC1 patients. Unfortunately, diagnosis of NPC1 disease is currently done by a set of consecutive tests, none of them being simultaneously simple and specific. Thus, it is critical to develop quick, inexpensive, and reliable diagnostic tests, which could be used in screening programs for NPC1 disease. In NPC1 patients, cholesterol oxidation products (oxysterols) are increased due to lysosomal accumulation of cholesterol and to cellular oxidative stress. Hence, oxysterol levels in blood can serve as specific biomarker for NPC1. In this project, we are interested in implementing a liquid chromatography mass spectrometry (LC-MS/MS) method for the analysis of oxysterols. Our goal is to obtain an easy and inexpensive diagnostic test that could be used for screening NPC1 patients. In addition, we are interested in understanding the potential role of oxysterols in other neurodegenerative disorders and in investigating the oxidative metabolism of cholesterol in more details.

Publications / Publikationen


Keywords / Suchbegriffe

Niemann-Pick C disease, lysosomal storage disorders, oxysterols, liquid chromatography, mass spectrometry, cholesterol metabolism.

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Funding Source(s) / Unterstützt durch

Private Sector (e.g. Industry)

In Collaboration with / In Zusammenarbeit mit

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Duration of Project / Projektdauer
May 2012 to Jan 2016