Washington University in St. Louis
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SENSITIVE AND ACCURATE MASS SPEC-BASED ASSAY FOR SIMULTANEOUS QUANTIFICATION OF GLYCOSYLSPHINGOSINE AND GALACTOSYLSPHINGOSINE

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Background

Gaucher and Krabbe disease are two types of lipid (fat) storage diseases that can result in progressive damage to the nervous system and other organ systems. Together, these diseases affect about 450,000 people worldwide. In both of these diseases, there is an undesirable accumulation of lipids. Blood levels of these lipids can be used for diagnosis or for monitoring the efficacy of treatments. However, while disease progression and symptoms are different for Gaucher and Krabbe disease, disease diagnosis using blood lipid levels are difficult because the lipids responsible for each disease are virtually indistinguishable using traditional methods. Glucosylsphingosine (Gaucher) and galactosylsphingosine (Krabbe) have the same molecular mass, both occur in low abundance, and are hard to separate for quantification.

Technology Summary

Dr. Ory's team has developed a method to simultaneously quantify the amount of each lipid. Using liquid chromatography-tandem mass spectrometry (LC-MS/MS) the team was able to break the lipids into different fragments for chemical analysis. This methodology allows for effective separation with higher sensitivity and has been successfully applied to measurement of these lipids in Krabbe animal models.

Key Advantages

- Enables specific and simultaneous quantification of glucosylsphingosine and galactosylsphingosine in serum and other tissues
- Rapid diagnosis and monitoring of diseases
- Direct quantification vs. indirect assessments currently used in the clinical tests for diagnosis
- Validated technology in a mouse model of Krabbe disease

Patent: Pending

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Dr. Ory is internationally recognized for his work on elucidating mechanisms underlying lipid

homeostasis, and the translation of the findings to develop biomarkers for prevention and treatment of human diseases.