Washington University in St. Louis
Office of Technology Management

NOVEL BILE ACIDS AS BIOMARKERS FOR EARLY DETECTION OF NIEMANN-PICK TYPE C DISEASE

Jiang, Xuntian, Ory, Daniel

Gill, John

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Technology Description

Researchers at Washington University in St. Louis have developed a set of novel bile acid biomarkers to screen, diagnose, and monitor the progression of Niemann-Pick type C (NPC) disease. Current testing for this disease takes an average of three months, is invasive (a skin biopsy) and is not generally performed until the appearance of symptoms. Additionally, there are no methods for screening in newborn babies.

This method is non-invasive, accurate, highly sensitive, and can be implemented in the screening of newborns. Thus, allowing for the early detection and diagnosis of NPC.

Stage of Research

The inventors have identified several novel bile acids that are effective biomarkers for NPC. They have developed an FDA-compliant clinical grade assay for quantifying these biomarkers using liquid chromatography-tandem mass spectrometry (LC-MS/MS) and compared this method to existing diagnostics.

Publications

- Jiang X and Ory, DS. (2016). Towards a new diagnostic standard for Niemann-Pick C. *EBioMedicine,* 4:18-9.
- Jiang X, Sidu R, Mydock L, Hsu F-F, … and Ory DS. (2016). Development of a Bile Acid-Based Newborn Screen for Niemann-Pick C Disease. *Sci Trans Med,* 8(337):337ra63.
- Jiang X, Sidhu R, Orsini JJ, Farhat NY, Porter FD, Berry-Kravis E, Schaffer JE, and Ory DS. (2019). Diagnosis of Niemann-Pick C1 by Measurement of Bile Acid Biomarkers in Archived Newborn Dried Blood Spots. *Mol Genet Metab*, 126:183-187.
- Sidhu R, Kell P, Dietzen DJ, Farhat NM, … and Jiang X. (2020). Application of a glycinated bile acid biomarker for diagnosis and assessment of response to treatment in Niemann-Pick disease type C1. *Mol Genet Metab*., in press.

Applications

- Diagnosis and monitoring of Niemann-Pick type C disease

Key Advantages

- **Detects NPC at birth, allowing for earlier detection and diagnosis**
- **Uses blood samples, a more non-invasive method, instead of skin biopsies to detect bile acids**
- **Method is specific for the disease, minimizing the occurrence of false positives**

- **Provides results faster than current methods**

Patents

Issued patent - US20170285015

- **Related Web Links –** Xuntian Jiang Profile