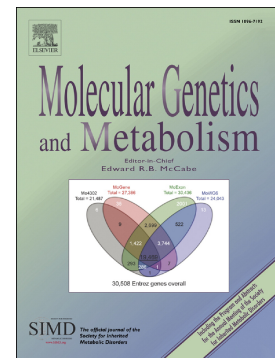


Accepted Manuscript

Urine oligosaccharide screening by MALDI-TOF for the identification of NGLY1 deficiency

Patricia L. Hall, Christina Lam, John J. Alexander, Ghazia Asif, Gerard T. Berry, Carlos Ferreira, Hudson H. Freeze, William A. Gahl, Kim K. Nickander, Jon D. Sharer, Caroline M. Watson, Lynne Wolfe, Kimiyo M. Raymond



PII: S1096-7192(18)30071-4
DOI: doi:[10.1016/j.ymgme.2018.03.002](https://doi.org/10.1016/j.ymgme.2018.03.002)
Reference: YMGME 6328
To appear in: *Molecular Genetics and Metabolism*
Received date: 14 February 2018
Revised date: 5 March 2018
Accepted date: 5 March 2018

Please cite this article as: Patricia L. Hall, Christina Lam, John J. Alexander, Ghazia Asif, Gerard T. Berry, Carlos Ferreira, Hudson H. Freeze, William A. Gahl, Kim K. Nickander, Jon D. Sharer, Caroline M. Watson, Lynne Wolfe, Kimiyo M. Raymond, Urine oligosaccharide screening by MALDI-TOF for the identification of NGLY1 deficiency. The address for the corresponding author was captured as affiliation for all authors. Please check if appropriate. Ymgme(2018), doi:[10.1016/j.ymgme.2018.03.002](https://doi.org/10.1016/j.ymgme.2018.03.002)

This is a PDF file of an unedited manuscript that has been accepted for publication. As a service to our customers we are providing this early version of the manuscript. The manuscript will undergo copyediting, typesetting, and review of the resulting proof before it is published in its final form. Please note that during the production process errors may be discovered which could affect the content, and all legal disclaimers that apply to the journal pertain.

Urine Oligosaccharide Screening by MALDI-TOF for the Identification of NGLY1 Deficiency

Patricia L. Hall¹, Christina Lam^{2,3,4}, John J. Alexander^{1,5}, Ghazia Asif¹, Gerard T. Berry⁶, Carlos Ferreira^{2,7}, Hudson H. Freeze⁸, William A. Gahl^{2,9,10}, Kim K. Nickander¹¹, Jon D. Sharer¹², Caroline M. Watson¹, Lynne Wolfe¹⁰, Kimiyo M. Raymond¹¹

¹EGL Genetic Diagnostics, LLC, Atlanta, GA

²Medical Genetics Branch, NHGRI, NIH, Bethesda, MD

³Division of Genetic Medicine, Department of Pediatrics, University of Washington School of Medicine, Seattle, WA

⁴Division of Genetic Medicine, Department of Pediatrics, Seattle Children's Hospital, Seattle, WA

⁵Department of Human Genetics, Emory University, Atlanta, GA

⁶The Manton Center for Orphan Disease Research, Division of Genetics and Genomics, Boston Children's Hospital, Harvard Medical School, Boston, MA, USA

⁷Division of Genetics and Metabolism, Children's National Medical Center, Washington, DC

⁸Human Genetics Program, Sanford Burnham Prebys Medical Discovery Institute, La Jolla, California

⁹Office of the Clinical Director, NHGRI, NIH, Bethesda, MD

¹⁰NIH Undiagnosed Diseases Program, Common Fund, Office of the Director, NIH, Bethesda, MD

¹¹Biochemical Genetics Laboratory, Department of Laboratory Medicine and Pathology Mayo Clinic College of Medicine, Rochester, MN

¹²Department of Genetics, University of Alabama Birmingham, Birmingham, AL

Corresponding Author:

Patricia L. Hall, Ph.D.

EGL Genetics

2460 Mountain Industrial Blvd.

Tucker, GA 30084

patriciahall@egl-eurofins.com

Download English Version:

<https://daneshyari.com/en/article/8343007>

Download Persian Version:

<https://daneshyari.com/article/8343007>

[Daneshyari.com](https://daneshyari.com)