

Accepted Manuscript

Exome Sequencing Analysis Reveals Variants in Primary Immunodeficiency Genes in Patients With Very Early Onset Inflammatory Bowel Disease

Judith R. Kelsen, MD, Noor Dawany, PhD, Christopher J. Moran, MD, Britt-Sabina Petersen, PhD, Mahdi Sarmady, PhD, Ariella Sasson, PhD, Helen Pauly-Hubbard, Alejandro Martinez, Kelly Maurer, Joanne Soong, Eric Rappaport, PhD, Andre Franke, MD, PhD, Andreas Keller, MD, PhD, Harland S. Winter, MD, Petar Mamula, MD, David Piccoli, MD, David Artis, PhD, Gregory F. Sonnenberg, PhD, Mark Daly, PhD, Kathleen E. Sullivan, MD, PhD, Robert N. Baldassano, MD, Marcella Devoto, PhD

PII: S0016-5085(15)00971-3
DOI: [10.1053/j.gastro.2015.07.006](https://doi.org/10.1053/j.gastro.2015.07.006)
Reference: YGAST 59887

To appear in: *Gastroenterology*
Accepted Date: 9 July 2015

Please cite this article as: Kelsen JR, Dawany N, Moran CJ, Petersen B-S, Sarmady M, Sasson A, Pauly-Hubbard H, Martinez A, Maurer K, Soong J, Rappaport E, Franke A, Keller A, Winter HS, Mamula P, Piccoli D, Artis D, Sonnenberg GF, Daly M, Sullivan KE, Baldassano RN, Devoto M, Exome Sequencing Analysis Reveals Variants in Primary Immunodeficiency Genes in Patients With Very Early Onset Inflammatory Bowel Disease, *Gastroenterology* (2015), doi: 10.1053/j.gastro.2015.07.006.

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.

All studies published in *Gastroenterology* are embargoed until 3PM ET of the day they are published as corrected proofs on-line. Studies cannot be publicized as accepted manuscripts or uncorrected proofs.



Exome Sequencing Analysis Reveals Variants in Primary Immunodeficiency Genes in Patients With Very Early Onset Inflammatory Bowel Disease

Short Title: WES analysis in patients with VEO-IBD

Judith R. Kelsen, MD¹, Noor Dawany, PhD², Christopher J. Moran, MD³, Britt-Sabina Petersen, PhD⁴, Mahdi Sarmady, PhD², Ariella Sasson, PhD², Helen Pauly-Hubbard¹, Alejandro Martinez¹, Kelly Maurer¹⁰, Joanne Soong⁵, Eric Rappaport, PhD⁶, Andre Franke, MD, PhD⁴, Andreas Keller, MD, PhD⁷, Harland S. Winter, MD³, Petar Mamula, MD¹, David Piccoli, MD¹, David Artis, PhD⁵, Gregory F. Sonnenberg, PhD⁵, Mark Daly, PhD^{8,9}, Kathleen E. Sullivan, MD, PhD¹⁰, Robert N. Baldassano, MD¹, Marcella Devoto, PhD¹¹

¹Division of Gastroenterology, Hepatology, and Nutrition, The Children's Hospital of Philadelphia,

²Department of Biomedical Health Informatics, The Children's Hospital of Philadelphia, ³Division of Pediatric Gastroenterology, Hepatology, & Nutrition, Massachusetts General Hospital for Children,

⁴Institute of Clinical Molecular Biology, Christian-Albrechts-University of Kiel, Germany, ⁵Joan and Sanford I. Weill Department of Medicine, Division of Gastroenterology and Hepatology, Department of Microbiology & Immunology, and The Jill Robert's Institute for Research in Inflammatory Bowel Disease, Weill Cornell Medical College, New York, New York, USA, ⁶Nucleic Acid/PCR Core, The Children's Hospital of Philadelphia, ⁷Department of Clinical Bioinformatics, Saarland University, Germany, ⁸Analytic and Translational Unit Center for Human Genetic Research Department of Medicine, Massachusetts General Hospital, ⁹The Broad Institute of MIT and Harvard, ¹⁰Division of Immunology and Allergy, The Children's Hospital of Philadelphia, ¹¹Division of Human Genetics, The Children's Hospital of Philadelphia, Department of Pediatrics, Department of Biostatistics and Epidemiology, Perelman School of Medicine, University of Pennsylvania; Department of Molecular Medicine, University Sapienza, Rome, Italy

Abbreviations: VEO-IBD (very early onset IBD), WES (whole exome sequencing), MAF (minor allele frequency), CVID (common variable immunodeficiency), EVS (Exome Variant Server), CADD (Combined Annotation Score), DHR (dihydrorhodamine)

Correspondence:

Judith R. Kelsen, MD
7NW, Division of Pediatric Gastroenterology
3400 Civic Center Blvd
The Children's Hospital of Philadelphia
Philadelphia, PA 19104
Tel: 215-590-3227
Fax: 215-590-5326
Kelsen@email.chop.edu

Disclosures: None of the authors have disclosures related to this manuscript.

Author Contributions: All authors contributed to this study, in particular:

Study concept and design: Kelsen, Dawany, Piccoli, Mamula, Artis, Sonnenberg, Baldassano, Sullivan, Devoto

Acquisition of data: Kelsen, Pauly-Hubbard, Martinez, Rappaport

Analysis and interpretation of data: Kelsen, Dawany, Petersen, Sarmady, Sasson, Rappaport, Artis, Sonnenberg, Sullivan, Baldassano, Devoto

Drafting of the manuscript: Kelsen, Dawany, Baldassano, Artis, Sonnenberg, Sullivan, Devoto

Critical Revision of Analysis of the manuscript for important intellectual content: Kelsen, Dawany, Moran, Sasson, Piccoli, Mamula, Artis, Sonnenberg, Daly, Sullivan, Baldassano, Devoto

Statistical analysis: Kelsen, Dawany, Sasson, Sarmady, Devoto

Technical or material support: Dawany, Maurer, Soong Sasson, Sarmady, Rappaport

Study Supervision: Kelsen, Baldassano, Devoto

Download English Version:

<https://daneshyari.com/en/article/6092345>

Download Persian Version:

<https://daneshyari.com/article/6092345>

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