

Comparison of fecal microflora in children with autism spectrum disorders and their 2 neurotypical siblings in the Simons Simplex Collection

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ABSTRACT

The Simons Foundation Autism Research Initiative (SFARI) has gathered a unique sample of children with autism spectrum disorder (ASD) and their neurotypical (NT) siblings in the Simons Simplex Collection (SSC). Sixty six simplex families in the SSC consented to participate in a study to compare the fecal microflora between ASD and neurotypical siblings of ASD probands aged 7-14. Functional gastrointestinal disorders (FGID) were assessed using ROME III process and problem behaviors were assessed using the Child Behavior Check List (CBCL). Targeted quantitative polymerase chain reaction (qPCR) assays were conducted on selected taxa, including *Sutterella*, *Bacteroidetes* and *Provetella*. Illumina sequencing of the V1-V2 and the V1-V3 regions of the 16S rDNA from fecal DNA to an average depth of 107,000 and 208,000 respectively. Thirty-two of 59 ASD children and 18 of 44 neurotypical siblings met ROME III criteria for an FGID. Functional constipation was more prevalent in ASD (17 of 59) compared to neurotypical siblings (6 of 44, $P = 0.035$). When ASD, FGID and first order ASD*FGID, were included as input variables in a negative binomial model, no significant associations were detected between ASD, FGID or first order ASD*FGID interactions and the relative abundance of *Sutterella*, *Bacteroidetes* or *Provetella* spp. by targeted qPCR or 16S rRNA sequence analysis. However, exploratory analysis of the 16S rRNA sequencing data identified several low abundance genera that were associated with ASD or first order ASD*FGID interactions, which will need to be confirmed in a larger sample set.

BIOGRAPHY

Dr. Ellen Li, MD, PhD, a gastroenterologist and research scientist, is Chief of the Division of Gastroenterology and Hepatology in the Department of Medicine at Stony Brook University School of Medicine. Dr. Li oversees the clinical, research and training of a Division that specializes in the prevention, diagnosis and treatment of diseases of the digestive tract and liver.

Dr. Li's major research interest is on defining the role of the gut microbiome in digestive diseases, particularly inflammatory bowel diseases, colon cancer, and functional GI disorders (e.g. irritable bowel syndrome). Since coming to Stony Brook in 2009, Ellen

has launched an interdepartmental Digestive Diseases Biobanking Core, with the generous support of the Simons Foundation, that will provide an infrastructure that encourages collaborations between clinicians and basic scientists in conducting clinical translational research. She is the author of 77 peer-reviewed articles and several chapters in the *Textbook of Gastroenterology*. Dr. Li participates in the training of medical students, residents, and fellows at the Stony Brook University School of Medicine.