The Clinical Rapid Genome Sequencing program implemented by CHGG has been expanded to the NYU Long Island Pediatric ICUs to provide rapid genomic diagnostics to critically ill children.

Gilad D. Evrony, MD, PhD, Assistant Prof. at CHGG and the Departments of Pediatrics, and Neuroscience and Physiology, was recently awarded the 2023 Pershing Square Sohn Prize for Young Investigators in Cancer Research. This is a prestigious annual award to empower High-Risk, High-Reward Cancer Research.
VEXAS (vacuoles, E1-ubiquitin-activating enzyme, X-linked, autoinflammatory, somatic) syndrome is a disease with rheumatologic and hematologic features caused by somatic variants in UBA1. Pathogenic variants are associated with a broad spectrum of clinical manifestations. Knowledge of prevalence, penetrance, and clinical characteristics of this disease have been limited by ascertainment biases based on known phenotypes. Dr. Beck and colleagues conducted a study to determine the prevalence of pathogenic variants in UBA1 and associated clinical manifestations in an unselected population. They provided an estimate of the prevalence and a description of the clinical manifestations of UBA1 variants associated with VEXAS syndrome.
Yu Zhang, PhD, presented her final MS Biomedical Informatics thesis project entitled “Construction of High-Resolution Epigenetic Maps of the Human Heart” on June 26 at the Vilcek Institute of Graduate Biomedical Sciences Biomedical Informatics Research Day. Yu generated and compared machine-learning-based high-resolution cardiac CRE maps for human adults and fetuses.
Congratulations to David Beck, MD, PhD, Assistant Prof. at CHGG and the Departments of Medicine and Biochemistry & Molecular Pharmacology for receiving the Department of Defense (DOD) award for his project “Beyond VEXAS and UBA1: Identification of Novel Drivers of MDS Autoimmune Syndromes.”

Congratulations to Samuel Magaziner, MD, PhD candidate in Dr. David Beck’s Lab, who was recently awarded the Fulbright Research Fellowship for his doctoral research project entitled: “Elucidating the mechanism of disease in VEXAS Syndrome.”

Congratulations to Gilad D. Evrony, MD, PhD, Assistant Prof. at CHGG and the Department of Pediatrics, Neuroscience, and Physiology for receiving a UG3 grant from the NIH Common Fund to develop a single-molecule sequencing technology for profiling single-strand damage and mismatches in DNA.
Hakhamanesh Mostafavi, PhD joins the CHGG.

Dr. Mostafavi will join the Center as Assistant Professor in the Division of Biostatistics, Department of Population Health.

Dr. Mostafavi is a computational biologist and population geneticist who received a PhD in Biological Sciences from Columbia University under the supervision of Molly Przeworski and conducted postdoctoral research with Jonathan Pritchard at Stanford University. His lab here will study human complex traits and diseases, with the aim of developing new models and methods to conceptualize their genetics, biology, clinical medicine and evolution.
CHGG Welcomes Two New Genetic Counselors

Mei-Kay Wong, MS, MPH, CGC, CPH

Mei-Kay is a new genetic counselor in the Beck Lab who will manage and investigate the molecular genetic basis of inflammatory diseases. Mei-Kay received her Master of Science in Genetic Counseling and Master of Public Health in Human Genetics from the University of Pittsburgh, Graduate School of Health.

Emma Mizrahi-Powell, MS, CGC

Emma is a new Research Genetic Counselor in the Evrony Lab who will manage the Undiagnosed Disease Program. Emma received her Master of Science in Genetic Counseling from Columbia University, Vagelos College of Physicians and Surgeons.
CHGG Welcomes New Members

Emilia Bianchini

Emilia is an entering Research Associate in the Evrony Lab. In her graduate research, she studied macrophage polarization and its implications for inflammation and osteoarthritis. Emilia received her Master’s degree in Biomedical Engineering from NYU.

Yu Zhang, PhD

Yu is a new Post-doctoral fellow in the Chakravarti Lab for the Center for Human Genetics and Genomics. Yu received her PhD from Rutgers Graduate School of Biomedical Sciences.

Emily McDonald

Emily came to the Center for Human Genetics and Genomics in May as a new Administrative Coordinator. Emily received her BS in Healthcare Management from Misericordia University, School of Business.

Vida Caraballo

Vida’s career at NYULH began in 2016 as a grants assistant for the Department of Plastic Surgery. She then moved to the Department of Population Health Sciences at Weill Medical College of Cornell University. Vida has returned home to NYULH as a Grants Manager under the PODs, where her main focus will be providing Pre and Post award services to the Center for Human Genetics & Genomics.

Gabe Grullon

Gabe is a recent Duke University graduate with a major in Neuroscience and a double minor in Chemistry and Biology. He will be joining the Chakravarti Lab as a Research Associate Technician. His work will be using mouse and cell culture models to study Hirschsprung's Disease and other complex genetic disorders.
CHGG Welcomes NTVs, SPIs, and Student Interns

Maysen Brato

Maysen is an NTV who was observing an “Autoimmune and Auto inflammatory Genetics Study” during her time with Genetic Counselor, Anna Cantor.

Athena Lam

Athena is a new Student Intern in the Beck Lab. Athena has an interest in genetics and is studying the molecular mechanism.

Odessa Muelbroek

Odessa was one of Dr. Carolyn Chapman’s high-school intern this year. Odessa focused on the “Ethical Issues in Human Genetics and Genomics.”

Phillip Seidell

Phillip was also one of Dr. Carolyn Chapman’s high-school interns this year. Phillip focused on the “Ethical Issues in Synthetic/Big DNA Biology.”

Lilah MacLowry

Lilah is a SPI under Dr. Aravinda Chakravarti. She is “Researching Comparative Genetics between Dogs and Humans and Assisting in DNA Extractions.”

Mudra Patel

Mudra was a SPI working with Or Yaacov on “building a tool that will allow Genetics Counselors to annotate VCF files based on their criteria of interest.” The goal is “to provide comfortable and convenient user experience for Genetic Counselors that will use this tool.”

Nhu Tran

Nhu is a SPI working together with Dr. Ryan Fine of the Chakravarti Lab. She is utilizing methods of synthetic biology and yeast genetics to explore a potential therapy for Hirschsprung disease.

Ustav Mesvani

Ustav Mesvani, a SPI working with Dr. David Beck, will be “Assisting with Molecular Biology Experiments and Learning Laboratory Techniques.”
Alumni: Joseph Tilghman, PhD

When did you join Chakravarti Lab, for how long, and in which position?
I joined the lab as a doctoral student in 2014, graduating in 2020.

What is your current position and when did you start?
I am a clinical genomics scientist at Invitae working on variant interpretation, reporting, and gene curation. I started in 2021.

How did you get to where you are today?
I graduated with an undergraduate degree in Biology from Washington and Lee University, where I became involved in research very early in my collegiate career. I increasingly became aware of the importance of genetics as a tool for understanding biology, leading me to my PhD in Human Genetics at the Johns Hopkins School of Medicine. I was a member of the Chakravarti Lab during the move from Hopkins to NYU, continuing to take part in the lab remotely from Baltimore as I completed my dissertation on the sequence analysis of familial neurodevelopmental disorders. The primary project from my dissertation to describe the molecular genetic anatomy of Hirschsprung disease was published in the New England Journal of Medicine in 2019.

How did your experience as a trainee in Chakravarti Lab help you along the way?
The Chakravarti lab embraces a very good balance with respect to the contributions of genetic variation to disease risk and the biological relevance of that variation, which helped me to develop a broad skillset and perspective with respect to understanding genetic disease at a population and individual level. I was constantly exposed to lab-mates and collaborators with varying backgrounds and views there was always a robust dialogue between these views.

What was the most difficult moment in your career and how were you able to bypass it and succeed?
The time during which I was working remotely following the move of the lab was a challenge with respect to working out any issues because there were added barriers to the regular feedback I was used to in the lab, but continued regular meetings with Aravinda were helpful, and learning how to maintain good working relationships across distance has proved to be very useful.

What advice would you give to trainees?
Maintain regular and frank communication with mentors and peers about difficulties you encounter in your work as a first step before trying to solve them independently.
On June 14th, the Center for Human Genetics and Genomics enjoyed an evening out together at a Sip and Paint. By the faculty and students attending this outing, a sense of community is built and moral is raised.

For the week of August 7th to the 11th, the Center of Human Genetics and Genomics will conduct a Research Adventure Course. CHGG faculty and senior trainees will provide an immersive introduction to human genetics and genomics research to incoming Vilcek graduate students. The center’s goal is to draw new students’ interest in human disease genetics throughout the campus but specifically highlight the many CHGG labs.
How to read a Professor's door

Closed
"I may or may not be on this continent."

Half-open
"I'm probably in a meeting."

Wide Open
"I just walked in to get a few things before I have to run to my next meeting."

Slightly ajar
"Proceed with caution."

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Feedback

We Want to Hear from You
We would like to feature your stories in the upcoming Newsletter. Please send us your Research Achievements: published articles, abstracts, presentations and awards. News, ideas, and feedback are always encouraged and can be sent to emily.mcdonald@nyulangone.org.

Who We Are
Center for Human Genetics & Genomics Faculty NYU Langone Health

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