

The CHGG* Newsletter



December 2024



*Center for Human Genetics & Genomics, NYULH

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CHGG 2024 Selected Publications

Evrony Lab - Murphy, Zachary R, et al.; Ultra-Rapid Droplet Digital PCR Enables Intraoperative Tumor Quantification. medRxiv, 2024

Evrony Lab - Groza C, et al. Pangenome graphs improve the analysis of structural variants in rare genetic diseases. Nature 15:657, 2024

Evrony Lab - Loh, Caitlin A, et al.; High-fidelity, large-scale targeted profiling of microsatellites. Genome Research 34:1008-1026, 2024

Beck Lab - Torreggiani S, et al. <u>Somatic mutations in autoinflammatory and autoimmune disease</u>. Nature Reviews Rheumatology 20:683-698, 2024

Beck Lab - Bisikirska Brygida, et al. <u>Melatonin receptor 1A variants as genetic cause of idiopathic osteoporosis</u>. Science translational medicine 16:eadj0085, 2024

Beck Lab - Kanagal-Shamanna R, et al. <u>Clonal Hematopoiesis, Inflammation, and Hematologic Malignancy.</u> Annual Review of Pathology 19:479-506, 2024

Beck Lab - Sirenko M, et al. Molecular and clinical presentation of UBA1-mutated myelodysplastic syndromes. Blood 144:1221-1229, 2024

Beck Lab - Oda H, et al. <u>Biallelic human SHARPIN loss of function induces autoinflammation and immunodeficiency.</u> Nature Immunology 25:764-777, 2024

Beck Lab - Kusne Y, et al. Venous and arterial thrombosis in patients with VEXAS syndrome. Blood 143:2190-2200, 2024

Beck Lab - Collins JC, et al. Shared and distinct mechanisms of UBA1 inactivation across different diseases. EMBO Journal 43:1919-1946, 2024

Beck Lab - Sébastien Küry et al. <u>Unveiling the crucial neuronal role of the proteasomal ATPase subunit gene *PSMC5* in neurodevelopmental proteasomopathies. *medRxiv*, 2024</u>

Beck Lab - Ospina Cardona D, et al. <u>Description of a novel splice site variant in UBA1 gene causing VEXAS syndrome</u>. *Rheumatology* (Oxford) 63:2897-2902, 2024



CHGG 2024 Selected Publications

Beck Lab - Czech M, et al. <u>Opportunistic Infections, Mortality Risk, and Prevention Strategies in Patients With Vacuoles, E1 Enzyme, X-Linked,</u> <u>Autoinflammatory, Somatic (VEXAS) Syndrome.</u> Open Forum Infectious Diseases 11:ofae405, 2024

Chakravarti Lab - Mukundan Attur, et al. Interleukin-1 Receptor Antagonist Gene (IL1RN) Variants Modulate the Cytokine Release Syndrome and Mortality of COVID-19. Journal of infectious diseases 229:1740-1749, 2024

Chakravarti Lab - Yaacov O, et al. Identification of the Molecular Components of Enhancer-Mediated Gene Expression Variation in Multiple <u>Tissues Regulating Blood Pressure</u>. Hypertension 81:1500-1510, 2024

Chakravarti Lab - Lee D, et al. <u>Tissue-specific and tissue-agnostic effects of genome sequence variation modulating blood pressure</u>. *Cell Reports* 42:113351, 2023

Chakravarti Lab - Gogineni E, et al. <u>Systematic Implementation of Effective Quality Assurance Processes for the Assessment of Radiation Target</u> <u>Volumes in Head and Neck Cancer.</u> Practical Radiation Oncology 14:e205-e213, 2024

Chakravarti Lab - Yaacov O, et al. <u>Identification of the Molecular Components of Enhancer-Mediated Gene Expression Variation in Multiple</u> <u>Tissues Regulating Blood Pressure</u>. Hypertension 81:1500-1510, 2024

Chakravarti Lab - Vincent E, et al. <u>Ret deficiency decreases neural crest progenitor proliferation and restricts fate potential during enteric nervous</u> system development. PNAS 120:e2211986120, 2023

Chatterjee Lab - Fries LE, et al. Variability in proliferative and migratory defects in Hirschsprung disease-associated *RET* pathogenic variants. *bioRxiv*, 2024



CHGG 2024 Featured Article



Shoutout to **Dr. Gilad Evrony** and his lab for work on HiDEFseq, which was featured in '<u>Spotlight on Science: NYU</u> <u>Grossman School of Medicine Research Digest</u>', the NIH director's blog, and published in <u>Nature</u>







DNA mismatch and damage patterns revealed by single-molecule sequencing

Mei-Hong Liu, et al.;

Nature 630:752–761, 2024 Read the article here







CHGG Academic Achievements



Ayana Sawai-Frantz, PhD NIH F32 Defining the role of Uba1 in bone marrow physiology and inflammation



Sumantra Chatterjee, PhD *NIH RO3* Computational identification of

Hirschsprung disease susceptibility genes



Gilad Evrony, MD, PhD *NIH RO1* Somatic mutation rates in healthy aging

Pershing Square Foundation Linking Brain Tumors to Their Embryonic and Fetal Lineages



David Beck, MD, PhD

Scleroderma Foundation Characterization of Somatic Mutations in Scleroderma



News & Views - Genome Health Analysis Program

Anna Cantor is partnering with co-director Claire Davis of Sarah Lawrence College to launch the novel Master of Science program in Genome Health Analysis (GHA). The GHA program aims "to meet the new and evolving needs of healthcare systems, professionals, and patients. Our goal is to bring to as many people as possible the benefits from the great advances that are being made in genomics."

After the program received its accreditation in December of 2023, the faculty and staff worked tirelessly to launch it. The official program launch took place in September 2024. This past April, **Anna** and **Dr. Chakravarti** joined the Administrative Leadership meeting to promote the GHA program internally.



Anna Cantor, MA, MS, CGC



Claire Davis, EdD, MS, CGC



<u>Click here</u> to watch Anna and Claire's recent podcast interview on DNA Today!

The podcast can also be found wherever you listen to others (Spotify, Apple Podcasts, etc.)



<u>Click here</u> to watch the April Administrative Leadership Meeting

Spotlight



Sam Magaziner's paper "Shared and distinct mechanisms of UBA1 inactivation across different diseases" made the cover of the Embo Journal in May.



Jenna Pucel, MS, CGC



Jenna and Emma passed their boards in March and became Certified Genetic Counselors.

Sumantra Chatterjee, PhD



Sumantra organized a Featured Symposium at the Annual Meeting of the American Society of Human Genetics in Denver this November. Further, Dr. Chatterjee has been invited to join the Mowat Wilson Syndrome Medical Advisory board. Lauren Fries and Sumantra also had Elsevier published their HSCR portion as part of their Life Science Collection. <u>Click here</u> to view the chapter/monograph. Sumantra was also awarded a Genetics Society of America Presidential Membership.



Rebecca Chubaryov

A graduate student in Aravinda Chakravarti's Lab was selected to moderate this session.



Shared and distinct mechanisms of UBA1 inactivation across different diseases

Collins JC, et al.

Embo Journal 43, 2024 Read the article here



CHGG Faculty Recruits



Hakhamanesh Mostafavi, PhD joined the Center for Human Genetics and Genomics in December 2023 as an Assistant Professor in the Department of Population Health. Dr. Mostafavi received his PhD in Biological Sciences from Columbia University in 2019, then continued to work as a Postdoctoral Scholar in the Department of Genetics at Stanford University. At CHGG, he focuses on computational and theoretical approaches to address fundamental questions about the biology of human traits and diseases. Welcome, Dr. Mostafavi!



Nadav Brandes, PhD received his PhD in Computational Biology from the Hebrew University of Jerusalem in 2021. He then joined the USCF School of Medicine as a Postdoctoral Scholar. Now, Dr. Brandes focuses on developing computational methods that leverage the power of AI and genomic language models to identify disease variants and genes to better understand complex genetic effects. Welcome, Dr. Brandes!

CHGG New Members



Theresa Scarabino – Program Manager Theresa joined the center in January and is coming from the Pandemic Response Lab. She has over 15 years of experience in the administration field. Welcome Theresa!



Lilin Huang – Associate Bioinformatics Programmer, Chakravarti Lab

Lilin got her MS in Computational Biology at Carnegie Mellon University. She has since joined the Chakravarti lab. **Welcome Lilin!**



Nisrine Jabara – Research Associate, Evrony Lab Nisrine graduated from Vassar College in May with a Bachelor of Arts in Biochemistry. She then joined the Evrony Lab in May. Welcome Nisrine!



Siddhi Bhutada – Research Associate, Chakravarti Lab Siddhi joined the Chakravarti Lab in July, and she is working on Cardiovascular diseases, focusing on Hypertension. Welcome Siddhi!



Jerome Hadjadj – Postdoc Fellow, Beck Lab Dr. Hadjadj came from the Internal Medicine outpatient clinic in Paris, France to join the Beck Lab in September. Welcome Jerome!



Allen Chen – Postdoc Fellow, Beck Lab Allen joined the Beck Lab in July and focuses on characterizing the systemic immune response in Sjogren's Disease using single cell profiling methods. Welcome Allen!



Akilah Williams – Administrative Assistant Akilah joined the center in August and is coming from the Mount Sinai Department of Dermatology. Welcome Akilah!



Angelika Tapia – Grants Manager

Angelika joined the center in November and has 21 years of finance experience, 15 years specifically in health care finance. Welcome Angelika!



CHGG Summer Students 2024



Sonali Campbell Chakravarti Lab



Stephy Chen Chakravarti Lab



Mehana Daftary Beck Lab



Baback Ghalamsiah Mostafavi Lab



Sophia Grullon Chakravarti Lab



Siri Kothapalli Chakravarti Lab



Alejandro Molina Beck Lab



Isagani Perez Beck Lab



Lauren Wilkes Chatterjee Lab



Tanvi Yadlapalli Chakravarti Lab

If you know someone who is interested in being a summer student at the CHGG, advise them to look at the <u>Beck</u>, <u>Evrony</u>, <u>Chakravarti</u>, <u>Brandes</u>, and <u>Mostafavi</u> lab websites.



GHA Master's Program Launch

In September, Anna Cantor, MA, MS, CGC, and Aravinda Chakravarti, PhD, hosted President Cristle Judd, PhD, co-director, Claire Davis, and faculty from Sarah Lawrence College to celebrate the official launch of the Genome Health Analysis program. CHGG is sending a big congratulations to **Anna Cantor** and a big welcome to the inaugural students, **Lisa Bura and Tvisha Bhatnagar!**











Genomics and Human Genetics: The New York Symposium

CHGG and Annual Reviews hosted 'Genomics and Human Genetics: The New York Symposium' in January 2024. Speakers from all around, including California and Paris, France, attended the symposium and gave talks on topics such as the 'Genome's Regulatory Code' and 'Telomereto-Telomere Assemblies'. With over 150 guests attending, the symposium was a great success! On the next page, those who presented posters are highlighted.

<u>Click here to view the program</u>



YU Grossman

Genomics and Human Genetics: The Symposium Poster Session

Members from CHGG labs, as well as students from Mount Sinai, joined the poster session, and presented and discussed their research to attendees.





EVENT

140 Years of Genetics...

On June 3rd, CHGG and the Institute of System Genetics jointly hosted a symposium to celebrate the 70th birthdays of Dr. Jef Boeke and Dr. Aravinda Chakravarti. Six noted geneticists from around the US came to deliver a lecture and join the celebrations. <u>Click here to view the program</u>





UCSC Genome Browser Workshop



Robert Kuhn, PhD, Associate Director of the Genome Browser, University of California Santa Cruz, visited NYU and taught ISG and CHGG students how the Genome Browser accesses "genome sequence data from a variety of vertebrate and invertebrate species and major model organisms".

<u>Click here to learn more about Dr. Kuhn</u> and the Genome Browser



ASHG Conference



Faculty and members from Chakravarti Lab attended the American Society of Human Genetics Annual Conference in Colorado this November. Members of the lab presented posters on their research, and Anna Cantor announced the GHA program.

Drs. Mostafavi and Chatterjee were chosen to give talks during the conference. Cheers to a successful trip!



NYU Grossman

School of Medicine





Make an appointment!



<u>Click here to schedule your NYU</u> Langone headshot appointment!

Wednesdays 9:20am-11:00am and 1:00pm-5:00pm

Thursdays 9:20am-12:00pm

Please email <u>media@nyulangone.org</u> if you require additional assistance.



Alumni – Mary Likhite





When did you join Beck Lab, for how long, and in which position?

I was a Research Associate in the Beck Lab from June 2022 until June 2024.

What is your current position now?

I am currently a first year PhD student at UMass Chan Medical School in the Graduate School of Biomedical Sciences.

Can you summarize some of the work you did in the Beck Lab?

I aided in setting up a variant calling pipeline to process the genomic data of patients David sees in clinic and who are referred to the lab. I also worked in the wet lab to process patient samples, verify different VEXAS mutations, and do some cell culture work for different projects.

How did your experience as a Research Associate in Beck Lab help you along the way?

I have found that the bioinformatics insights and techniques I learned in the Beck Lab are indispensable and give me a huge edge over other members of my cohort in making insights in different papers, exercises, and lectures. Having wet lab experience in addition (beyond an undergraduate lab experience) has also really helped me get my foot in the door with different PIs for potential rotations. A thing I've noticed about having worked in a research lab in general before going back to school is that I'm a lot more comfortable figuring things out on my own than others in my cohort. Some people complain during their rotations that they are not explicitly trained on every experiment they need to do, while I am a lot more at ease researching the best way to carry things out with the resources, I have available. Not only can I get the work done more efficiently, but I understand more about the process than just mindlessly following a protocol. Part of innovation and novel findings is understanding that there isn't a guidebook for everything.

Did you ever consider an alternative career path?

The master's program I pursued was primarily geared at getting people into industry. I had originally intended to become a research associate or technician at a biotech company. I got the opportunity to work in David's lab and re-routed my plans to go back to academia, still considering going to biotech after my time in the Beck Lab. Working in the Beck Lab helped me figure out where I wanted my career to go.

What advice would you give to other RAs?

Go to every lecture, seminar, and event that you can fit into your schedule. This period should be about pursing cutting edge research, learning about different scientific fields, and making connections with other scientists. Even if you think you know where you want to end up, try to experience a wide variety of different subjects. You never know what's going to pique your interest.

Any additional comments and related stories you might want to add?

It would absolutely be when we took a department outing to Bowlero. I figured out if I threw the bowling ball as hard as I possibly could I could send pins flying, and I spent three games absolutely annihilating my opponents.

The Comic Strip







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
- Awards

Who Are We?

Center for Human Genetics & Genomics Faculty NYU Langone Health

CHGG Home Page

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News, ideas, and feedback are always encouraged and can be sent to <u>emily.mcdonald@nyulangone.org</u>.