

CHGG Chronicles

November 2023



Center for Human Genetics and Genomics



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Featured Article

“Ret deficiency decreases neural crest progenitor proliferation and restricts fate potential during enteric nervous system development.”

Elizabeth Vincent, Sumantra Chatterjee, Gabrielle H. Cannon, Dallas Auer, Holly Ross, Aravinda Chakravarti, Loyal A. Goff
Proc Natl Acad Sci USA 2023 Aug 22;120(34):e2211986120. doi: 10.1073/pnas.2211986120. Epub 2023 Aug 16.

In their latest publication in the Proceedings of the National Academy of Science (PNAS), the Aravinda Chakravarti and Sumantra Chatterjee labs, in collaboration with Loyal Goff’s lab at John Hopkins University, describe how loss of function of Ret, a critical gene associated with Hirschsprung disease (HSCR), results in reduced proliferative capacity leading to a loss of predominantly inhibitory motor neurons.



Aravinda Chakravarti, PhD

Sumantra Chatterjee, PhD

Academic Achievements



David Beck, MD, PhD, receives another grant

David Beck received a grant from the Gabrielle's Angel Foundation for Cancer Research for his research on **“Modulating UBA1 to Treat Myelodysplastic Syndrome.”** Congratulations, David!



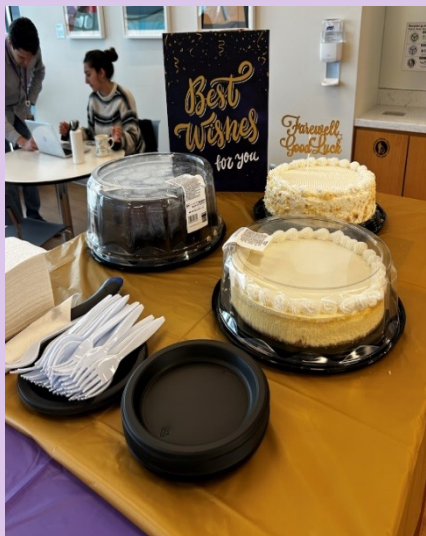
NIH awards F30 to Sam Magaziner

Sam Magaziner, MD, PhD student in the Beck lab, received the Ruth L. Kirschstein National Research Service Award (NRSA) F30 Grant from NIH/NHLBI to pursue his research project **“Elucidation of the Mechanism of Disease in VEXAS Syndrome.”** Congratulations, Sam!

Dr. Carolyn Chapman's Departure



Carolyn R. Chapman, PhD



We want to thank Dr. Carolyn Chapman for her contributions to the CHGG and wish her all the best as she starts a new position with the Multi-Regional Clinical Trials Center of Brigham and Women's Hospital and Harvard (MRCT Center). Carolyn has served as Faculty in the CHGG with a primary appointment as Research Assistant Professor in the Department of Population Health (Division of Medical Ethics). She has served as Director of Research Ethics Education in the Translational Research Education and Careers (TREC) Unit of the NYU Clinical & Translational Science Institute (CTSI) since June 2020. The CHGG celebrated Dr. Chapman's work on September 27th. Please wish her the very best success moving forward!



Jessica DiMaio, MHA

Jessica DiMaio's Departure

CHGG would like to give immense thanks to Jessica DiMaio who has recently stepped down from her position. Jessica was one of our first employees, whom we recruited in 2018. She performed with aplomb in any of the roles we wanted her to manage, putting in a great deal of work and sweat to make the CHGG a success. Her dedication towards the administrative team and the faculty is notable. Jessica graduated from Towson University with a Bachelor of Science degree in 2014, and continued her journey to Hofstra University where she graduated with a Masters in Health Administration degree in 2016. She started her career with the CHGG as an Administrative Coordinator. We wish Jessica the very best of luck in the future!



Sam, Gabe, and Emily

Fire Lab and Safety Team New Volunteers

CHGG Fire Lab and Safety Team (FLST) welcomes three new volunteers: **Sam Magaziner, Gabe Grullon, and Emily McDonald**. They are in charge of guiding everyone on SB 8th floor to safety and alerting the correct department, in case of emergency. On October 26th, they took on their first training session. Thanks Sam, Gabe, and Emily for all your help in guiding CHGG to safety!

Spotlight: Lilah MacLowry



Lilah MacLowry

Lilah MacLowry, an undergraduate from Georgetown University, shares her summer experience working as a Sponsored Project Individual (SPI) in the Chakravarti lab. She learnt methods for detecting and confirming *de novo* mutations in Hirschsprung disease alongside Dr. Hanna Berk-Rauch and graduate student Mingzhou Fu.

“I primarily focused on validating ultra-rare *de novo* mutations in Hirschsprung disease using Sanger sequencing. Through this project, I learned how to perform PCR, gel electrophoresis, gel extraction/purification, and how to analyze sequencing data. Outside of specific methodologies, I also learned about the process of troubleshooting these protocols and the thought process behind each experiment. This was very valuable to me because understanding the foundations of how to approach a challenge will be applicable to any further research I participate in. In my second project, I learned cell culture techniques, RNA extraction, and RT-PCR in order to investigate sex differences in Autism. Finally, I learned the comparative genomics in animal models through a literature review. Overall, everyone in the lab was extremely open and willing to help facilitate my learning and I greatly appreciate the mentorship I received.”

Spotlight: Sumantra Chatterjee, PhD



Sumantra Chatterjee, PhD

On October 20th, 2023, **Sumantra Chatterjee, PhD**, gave a seminar on the **“Development and Disorders of the Enteric Nervous System”** at the CGD Monthly Research Talks hosted by Marcin Imielinski, MD, PhD.

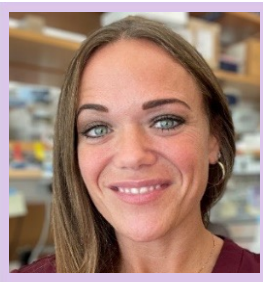
“I discussed my lab’s work uncovering the genetic and cellular basis of the development of the enteric nervous system and its disorders. It was a well-attended seminar by trainees and faculties and was followed by interesting and informative discussion on the projects and their implications for the broader NYU community.”

CHGG Welcomes New Members



Amoolya Srinivasa
Evrony Lab

Amoolya joined the Evrony Lab as a **Bioinformatics Programmer**. She received her MS Bioinformatics degree this year from Northeastern University. She will be collaborating with Dr. Evrony to help unravel new biological insights for rare diseases.



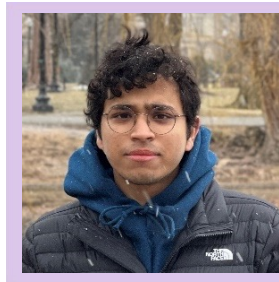
Debbie Hernandez
Beck Lab

Debbie has joined the Beck Lab to be the **Lab Manager** after 25 years in the Reinberg Lab at NYU Langone Health.



Jenna Pucel
Chakravarti Lab

Jenna is a **Research Genetic Counselor** in the Chakravarti Lab. She received her MS in Genetic Counseling from the MGH Institute of Health Professions. She will be involved in study participant outreach, recruitment, and consenting for HSCR studies.



Manas Dhanuka
Chakravarti Lab

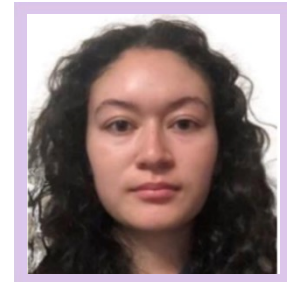
Manas is a Biotechnology Engineer with a Master's in Bioinformatics from Boston University. He is the new **Bioinformatics Programmer** for both the Aravinda Chakravarti Lab and the Shukti Chakravarti Lab.

Manas is working on managing, optimizing, structuring, and storing data for the Aravinda Chakravarti lab along with working on the Autism Project. For the Shukti Chakravarti Lab, he will be assisting in the computational analysis of RNA seq data.



Michelle Rivera Lomeli
Chakravarti lab

Michelle is a recent graduate of University of California Los Angeles, where she pursued a B.S. in Neuroscience. Michelle has since forth joined the Chakravarti Lab as a **Research Associate Technician**. She will use cellular and mouse models to study functional genomics in neurodevelopmental diseases.



Sachiko Keane
Beck Lab

Sachiko has an undergraduate degree in Neuroscience and a concentration in statistics from Columbia University. She is currently a **Research Associate** in the Beck Lab, establishing the autoimmune inflammatory disease VEXAS in mouse models.

CHGG Welcomes Three New Rotation Students



Amy Kwan
Beck Lab

Amy Kwan graduated from Washington University in Saint Louis, studying Computational Biology and Genomics. She is currently rotating in the lab of Dr. David Beck to create a mouse bone-marrow-derived macrophage model of VEXAS.



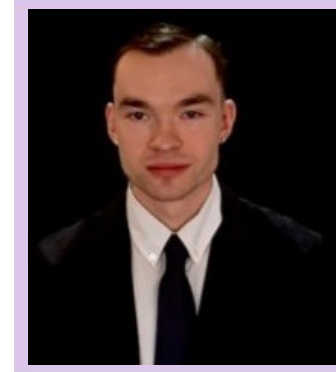
Jacob Stimes
Evrony Lab

Jacob Stimes gained his BS in software engineering at Iowa State University. He is now a first year PhD student rotating in the lab of Dr. Gilad Evrony, studying the genetics of brain tumors.



Amy Wang
Beck Lab

Amy Wang graduated from UC Santa Barbara, majoring in cell and developmental biology. She will be rotating in the lab of Dr. David Beck to gain insight into the mechanism of VEXAS syndrome.



Avery Dunn
Chakravarti Lab

Avery is apart of the Chakravarti lab focusing on computational gene regulation analysis. He is building enhancer maps for genes related to Hirschsprung's disease such as RET through analysis of human fetal ATAC-seq data.

Alumni: Mei Hong Liu



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

I joined the Evrony Lab in June 2020 as a research associate. I was there for two years before leaving to start medical school.

What is your current status now?

I am currently a second year medical student at the Albert Einstein College of Medicine.

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

As a research associate, I worked on several independent projects with guidance from Dr. Gilad Evrony to develop novel genomic technologies that would enable us to study somatic mutations in greater depth. One of these projects was developing a new ultra-high fidelity sequencing method called Hairpin Duplex Enhanced Fidelity Sequencing (HiDEF-seq), that allowed us to detect single-stranded mutations. We used HiDEF-seq to sequence a variety of samples, including samples from individuals with cancer-predisposition syndromes, to define the first single-strand mismatch and damage signatures. If you would like to learn more about HiDEF-seq, the paper is currently published on BioRxiv:

<https://www.biorxiv.org/content/10.1101/2023.02.19.526140v1.full.pdf>

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

My experience in the Evrony Lab and the skills I have acquired has been tremendously helpful to me now as a medical student. The knowledge and exposure I had to molecular biology, human genetics, and genomics has helped me in better understanding the disease processes I learn about in my classes. The skills I have acquired were not only transferable to basic science research in other disciplines but also unfamiliar forms of research such as clinical or qualitative research that I am currently involved in.

Did you ever consider an alternative career path?

When I joined the Evrony Lab, I was considering which path to choose out, MD, MD/PhD, or PhD. Dr. Evrony's lab was the perfect place for me to explore these paths since the lab is composed of a mixture of people with MD/PhDs and PhDs. Working in the lab, shadowing the Undiagnosed Diseases Program, and speaking with Dr. Evrony, graduate students, and post-doc in the lab gave me a pretty good picture of what a career with an MD/PhD could look like. I ultimately decided to pursue a MD degree and supplement my training with different research opportunities I could engage in as a student.

What advice would you give to other Research Associates?

My advice for other research associates is to ask for help, learn from your other lab members, and lean on them for support. My lab members have not only helped me with my research but have also helped and supported me when I was applying to medical school. Shout out to the Evrony Lab members, you are all amazing and I couldn't have done it without you all!

CHGG interviews **Mei Hong Liu**, a former Research Associate in the Evrony Lab. Mei shares her experience and advice from her time here at CHGG.

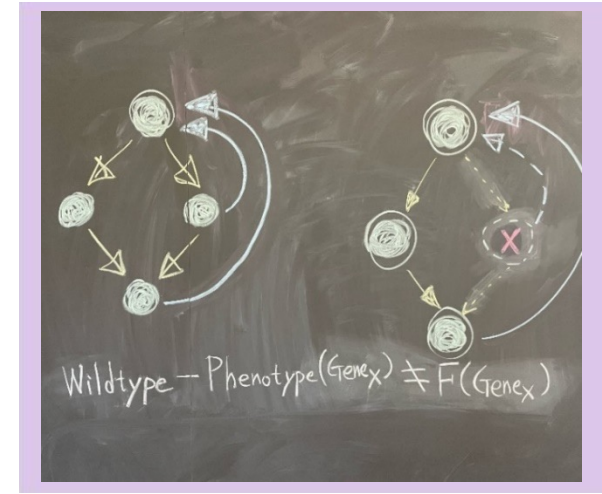
CHGG Seminars



Anastasia Wise, PhD



Kym Boycott, MD, PhD, FRCPC, FCCMG



Gaudenz Danuser, PhD

Anastasia Wise, PhD, is the Director of Scientific Return to Participants and Impact for the **All of Us Research Program**, and leads the responsible return of genomic results to participants. Dr. Wise spoke about the *All of us* Research Program at the Center for Human Genetic and Genomics “Genetic Medicine Colloquium” on September 26th, hosted by Dr. David Beck.

Kym Boycott, MD, PhD, FRCPC, FCCMG, is the Chair in the Department of Genetics at the Children’s Hospital of Eastern Ontario.

Dr. Boycott discussed the **functional studies on the discovery of new causes of rare diseases** at the Center for Human Genetics and Genomics “Genetic Medicine Colloquium” on October 24th, hosted by Dr. Gilad Evrony.

On November 14th, 2023, Gaudenz Danuser, PhD, will present his seminar on **“Causal inference in complex molecular systems”**, hosted by Alex Mogliner, PhD, in affiliation with NYU Grossman School of Medicine, NYU Courant Institute of Mathematical Sciences, and the Tandon School of Engineering and School of Arts and Sciences at the Computational Biology and Medicine Colloquium.

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CHGG Seminars



Jack Humphrey, PhD

Instructor in the Department of Neuroscience at Mount Sinai School of Medicine, Jack Humphrey, PhD, presented his seminar on **“Functional Genomics across neurodegenerative disease uncovers novel genes, pathways, and cell-types”** for NYULH on October 26th, which was hosted by Aravinda Chakravarti, PhD, and Steven Galetta, MD.



Hanna Berk-Rauch, PhD

Hanna Berk-Rauch, PhD, senior research scientist in the Chakravarti lab, presented at the ISG Nano Seminar on October 26th about the **“Genetic Analysis of Sex Difference in Autism”**, which was hosted by Aravinda Chakravarti, PhD.



Anne Goriely, PhD

Dr. Anne Goriely, Professor of Human Genetics, Radcliffe Department of Medicine, University of Oxford, visited the Center for Human Genetics and Genomics and the Department of Biochemistry and Molecular Pharmacology on October 30th to discuss her thoughts on **“Ageing men, their selfish testes, new mutations and human disease”**.

CHGG Events



Vilcek Students with CHGG faculty
August 7th – 11th, 2023

From August 7th to August 11th, the Center for Human Genetics and Genomics welcomed incoming Vilcek graduate students to **Research Adventure Week**. During the week, the students met with CHGG faculty, senior trainees, and genetic counselors to explore human disease genetics. They delve into topics such as “Genomic Analyses of Disease Genes” and “Mouse and Other Models of Human Disease Genes.” On that Friday, the students enjoyed a happy hour event with CHGG faculty, post-docs, and students.



ASHG
November 1st – 5th, 2023

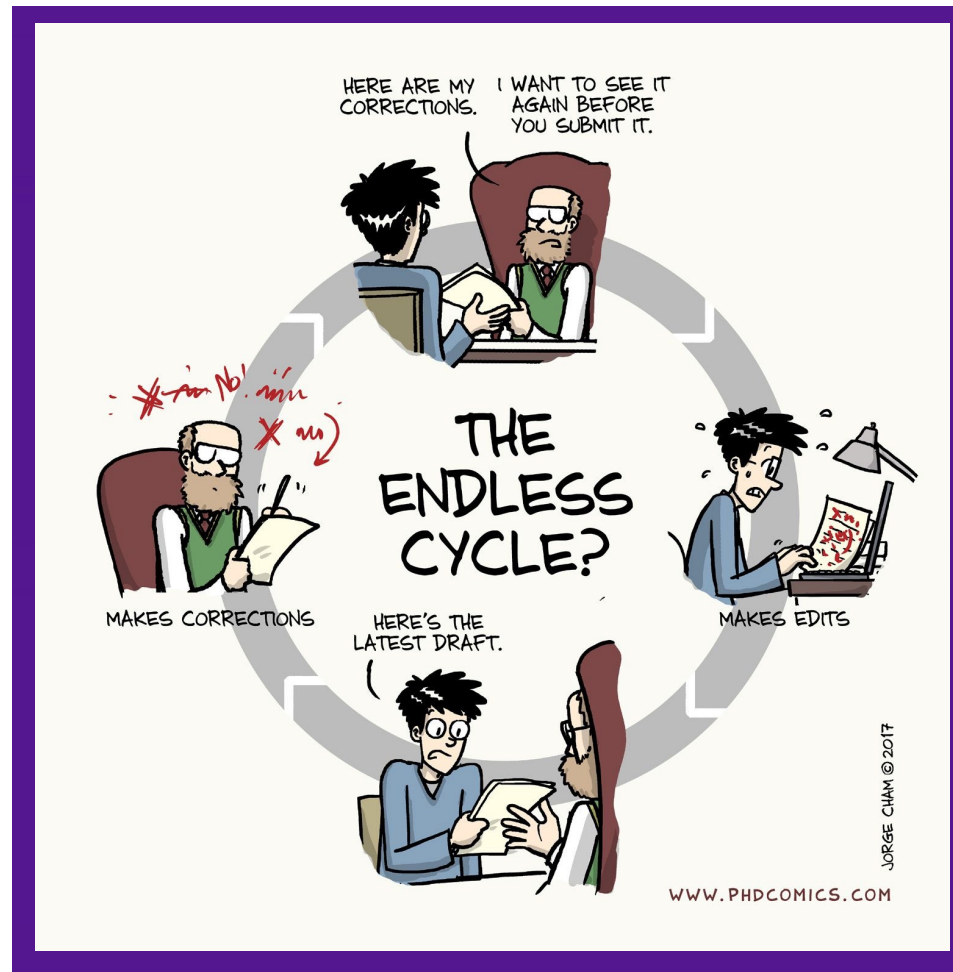
Members of CHGG will be attending the American Society of Human Genetics annual meeting on November 1st through the 5th. The members attending are **Hanna Berk-Rauch, Kristen Ayers, Joydeep Mitra, Karan Singh, Yu Zhang, and Brecca Miller. Sumantra Chatterjee, Ryan Fine, Rebecca Chubaryov, and Druha Karunakaran** will present their research in a poster session.



CHGG Holiday Party
December 7th, 2023

On December 7th, the Center for Human Genetics and Genomics will enjoy their well deserved holiday party. The party will be held in the Science Building 8th floor pantry area, where everyone will socialize and celebrate the upcoming holidays. The Center for Human Genetics and Genomics hopes everyone enjoys their holiday celebrations!

Comic



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 Are We?

Center for Human Genetics & Genomics Faculty NYU Langone Health

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