# Rahul Gupta

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## Education

2017–Present **MD Candidate**, *Harvard Medical School*, Boston, MA
Harvard-MIT Program in Health Sciences and Technology (HST)

2019–2023 **Doctor of Philosophy**, *Harvard Medical School*, Boston, MA
Biological and Biomedical Sciences, Program in Genetics and Genomics
Co-advised by Drs. Vamsi Mootha & Benjamin Neale

2012–2016 **Bachelor of Science in Engineering**, *University of Pennsylvania*, Philadelphia, PA, Summa Cum Laude
School of Engineering and Applied Science (SEAS) GPA: 3.96/4.00
**Majors:** Chemical & Biomolecular Engineering, Biology; **Minor:** Chemistry MCAT Score: 524/528

## Selected publications and preprints

(* indicates co-first author)

2024 Karczewski, KJ*, **Gupta, R***, Kanai, M*, et al. Pan-UK Biobank GWAS improves discovery, analysis of genetic architecture, and resolution into ancestry-enriched effects. medRxiv 2024.

2023 **Gupta, R**, Kanai, M, Durham, TJ, et al. Nuclear genetic control of mtDNA copy number and heteroplasmy in humans. Nature 2023.

2023 **Gupta, R**, Sharma, R, Gijavanekar, C., et al. A metabolite score that quantifies mitochondrial dysfunction at population scale. Manuscript in preparation.

2021 **Gupta, R**, Karczewski, KJ, Howrigan, D, et al. Human genetic analyses of organelles highlight the nucleus, but not the mitochondrion, in age-related trait heritability. eLife 2021.

2020 Rath S*, Sharma, R*, **Gupta, R***, et al. MitoCarta3.0: an updated mitochondrial proteome now with sub-organelle localization and pathway annotations. Nucleic Acids Res. 2020.

2018 Jha, P*, McDevitt, MT*, **Gupta, R***, et al. Systems Analyses Reveal Physiological Roles and Genetic Regulators of Liver Lipid Species. Cell Systems 2018

2017 **Gupta, R***, Lan, M*, Mojsilovic-Petrovic, J, et al. The Proline/Arginine Dipeptide from Hexanucleotide Repeat Expanded C9ORF72 Inhibits the Proteasome. eNeuro 2017

## PhD Dissertation

2019–2023 **Harvard Program in Biological and Biomedical Sciences (BBS), Genetics and Genomics Concentration**, *Graduate student*, Boston MA

Advisors: Vamsi K. Mootha, MD, Benjamin M. Neale, PhD

Title: Nuclear genetic control of mitochondrial function and its contribution to human disease: insights at biobank scale

- Used statistical genetics at scale to gain basic biological insights about mitochondria and metabolism
- Created metabolomic models in patients with rare disorders to gain insights into common disease
- Developed methods to extract mtDNA endophenotypes using whole-genome sequencing data
- Analyzed the contributions of variation in mitochondria-localizing genes to disease heritability
- Assessed phenotype heritability across diverse populations in UK Biobank
- Developed large-scale computational pipelines using Google Cloud Platform

## Consulting experience

2023–Present **Marea Theraputics**

Contact: Beryl Cummings, PhD

- Created tools to analyze large-scale genotype and phenotype data to prioritize targets
- Developed pipelines to produce disease-specific cohorts for genetic discovery

## Honors and fellowships

2021–2025 **NIH Ruth L. Kirschstein National Research Service Award (NRSA F30)**, *National Institute on Aging*

2021 **Paul and Daisy Soros Fellowship for New Americans — Finalist**

2021 **DoD NDSEG Fellowship Program — Waitlist**

2019 **Outstanding Student Poster Presentation**, *New England Society of General Internal Medicine*

2016 **Rose Award**, *UPenn Center for Undergraduate Research and Fellowships*

2016 **Neysa Cristol Adams Prize in Biology – Honorable Mention**, *UPenn Department of Biology*

2016 **Hugo Otto Wolf Memorial Prize**, *UPenn School of Engineering and Applied Science*

2016 **A. Norman Hixson Prize in Chemical Engineering**, *UPenn Department of Chemical and Biomolecular Engineering, CBE*

2016 **Senior Delaware Valley Section Award**, *Delaware Valley Section of the American Institute of Chemical Engineers, AIChE*

2016 **President's Innovation Prize – Semifinalist**, *UPenn*, Diagnosis of malaria in low-resource settings with AmpliTest

2015 **Donald F. & Mildred Topp Othmer Scholarship Award**, *AIChE National*

2015 **Junior Delaware Valley Section Award**, *Delaware Valley Section of AIChE*

2014 **Donald F. Othmer Sophomore Academic Excellence Award**, *UPenn CBE*

2014–2016 **Tau Beta Pi**, *Pennsylvania Delta Chapter*

2012–2014, 2015–2016 **Dean's List**, *UPenn School of Engineering and Applied Science*

## Other publications and preprints

(* indicates co-first author)

2024 Rogers, RS, Sharma, R, Shah, HS, Skinner, OS, Guo, A, Panda, A, **Gupta, R**, et al. Circulating N-lactoyl-amino acids and N-formyl-methionine reflect mitochondrial dysfunction and predict mortality in septic shock. Metabolomics 2024.

2024 Rath, S, **Gupta, R**, Todres, E, Wang, H, et al. Mitochondrial genome copy number variation across tissues in mice and humans. Manuscript under peer review.

2023 Wharrie, S, Yang, Z, Raj, V, Monti, R, **Gupta, R**, et al. HAPNEST: efficient, large-scale generation and evaluation of synthetic datasets for genotypes and phenotypes. Bioinformatics 2024.

2023 Majara, L, Kalungi, A, Koen, N, Tsuo, K, Wang, Y, **Gupta, R**, et al. Low and differential polygenic score generalizability among African populations due largely to genetic diversity. HGG Adv. 2023.

2022 Tsuo K, Zhou, W, Wang, Y, Kanai, M, Namba, S, **Gupta, R**, et al. Multi-ancestry meta-analysis of asthma identifies novel associations and highlights the value of increased power and diversity. Cell Genomics 2022.

2021 The COVID-19 Host Genetics Initiative. Mapping the human genetic architecture of COVID-19. Nature 2023.

2018 Jha, P, McDevitt, MT, Halilbasic, E, Williams, EG, Quiros, PM, Gariani, K, Sleiman, MB, **Gupta, R**, et al. Genetic Regulation of Plasma Lipid Species and Their Association with Metabolic Phenotypes. Cell Syst. 2018

## Talks

2025 **Gupta, R**, (2025). Uncovering mechanisms driving mtDNA variation across 750,000 people. Freedom Together Foundation Annual Meeting. Oral presentation, plenary session.

2024 **Gupta, R**, (2024). Biobank-scale analysis of mtDNA copy number and heteroplasmy. Mass. General Hospital Center for Genomic Medicine Seminar Series. Oral presentation.

2024 **Gupta, R**, (2024). Nuclear genetic control of mtDNA homeostasis revealed from >250,000 diverse human genomes. *All of Us* Office Hours Researcher Presentation, Invited oral presentation.

2023 **Gupta, R**, (2023). Nuclear genetic control of mtDNA homeostasis revealed from >250,000 human genomes. Keystone Meeting; Mitochondrial Dysfunction: From Ultra-rare Diseases To Aging, Oral presentation, plenary session.

2023 **Gupta, R**, (2023). Nuclear genetic control of mtDNA homeostasis revealed from >250,000 human genomes. EUROMIT. Invited oral presentation.

2023 **Gupta, R**, (2023). Nuclear genetic control of mtDNA homeostasis revealed from >250,000 human genomes. Broad Institute Board of Scientific Counselors Meeting. Oral presentation.

2022 **Gupta, R**, (2022). The genetic determinants of mitochondrial function and aging in humans. Howard Hughes Medical Institute Deconstructing and Decoding the Genome Conference, Virtual.

2022 **Gupta, R**, (2022). Assessing mitochondrial dysfunction at biobank scale. Harvard Program in Genetics and Genomics Seminar. Oral presentation.

2021 **Gupta, R**, (2021). Quantifying mitochondrial dysfunction at biobank scale. Mass. General Hospital Center for Genomic Medicine Seminar Series. Oral presentation.

## Posters

2024 **Gupta, R**, Chau, G, Durham, T, et al. (2024). Pervasive human mitochondrial-nuclear interactions and their implications for disease identified across >600,000 individuals. American Society of Human Genetics.

2022 **Gupta, R**, Sharma, R, Surendran, P, et al. (2022). Quantifying mitochondrial dysfunction at biobank scale using insights from rare disease. American Society of Human Genetics.

2022 **Gupta, R**, Karczewski, KJ, Howrigan, D, Neale, BM, Mootha, VK (2022). Organelles and aging: a human genetic approach. Harvard Program in Genetics and Genomics Symposium.

2021 **Gupta, R**, Karczewski, KJ, Howrigan, D, Neale, BM, Mootha, VK (2021). Organelles and aging: a human genetic approach. American Society of Human Genetics. Virtual.

2021 **Gupta, R**, Karczewski, KJ, Howrigan, D, Neale, BM, Mootha, VK (2021). Human genetic analyses of organelles highlight the nucleus, but not the mitochondrion, in age-related trait heritability. Mass. General Hospital Scientific Advisory Board Meeting. Virtual.

2019 **Gupta, R**, McGirr, S, Trainor, A (2019). An acute presentation of hereditary pancreatitis in a 24-year-old. New England Regional Society of General Internal Medicine.

2019 **Gupta, R**, Calvo S, Aguet F, Mootha VK (2019). Human Genetics of Mitochondrial DNA Copy Number Variation. Harvard Medical School Soma Weiss Student Research Day; Boston, MA.

2017 **Gupta, R**, McDevitt M, Jha P, et al. (2017). Identification and Bioinformatic Network Analysis of Lipid Quantitative Trait Loci. Morgridge Scientific Advisory Board Meeting; Madison, WI.

2016 **Gupta, R**, Lan M, Mojsilovic-Petrovic J, et al. (2016). The Proline/Arginine Dipeptide from C9orf72 RAN Translation Leads to Cellular Degradation Pathway Dysfunction. Penn Symposium on Undergraduate Research in Biology; Philadelphia, PA.

## Previous research and laboratory experience

2018–2019 **Broad Institute of MIT and Harvard**, *MD Research Assistantship*, Cambridge MA

Advisor: Vamsi K. Mootha, MD

Topic: Characterizing human variation in mtDNA copy number (mtCN) across tissues and individuals

- Measured mtCN across ≈10,000 human samples
- Systematically assessed of genetic and transcriptomic correlates to mtCN across tissues

2016–2017 **Morgridge Institute for Research**, *Research assistant*, Madison WI

Advisor: David Pagliarini, PhD

Topic: Leveraging systems genetics strategies to understand the link between liver and serum lipid species and clinical and molecular traits

- Created a pipeline to nominate genes from quantitative trait loci for lipidomics in mice
- Integrated of genetic, transcriptomic, proteomic, and metabolomic data

2014–2016 **Children's Hospital of Philadelphia**, *Undergraduate researcher*, Philadelphia PA

Advisor: Robert Kalb, MD

Topic: Proteasome-mediated toxicity of dipeptide products from C9ORF72 hexanucleotide repeat expansion (HRE) in amyotrophic lateral sclerosis

- Used of molecular techniques to probe the impact of peptide products from C9ORF72 HRE

- Assessed of proteasomal and autophagic flux impairment in C9ORF72 HRE

2015–2016 **Univ. of Pennsylvania Departments of Chemical Engineering & Bioengineering**, *Interdisciplinary senior design project*, Philadelphia PA

Advisor: Haim Bau, PhD

Topic: Diagnosis of malaria in low-resource settings with AmpliTest

- Created an electricity-free diagnostic platform for P. falciparum using isothermal amplification
- Generated a low-cost prototype performed validation using 3D-printing and computer-aided design

2013 **Cytec Industries (now SOLVAY) Analytical Laboratories**, *Internship*, Stamford, CT

Manager: Min Wang, PhD

- Developed methods for HPLC, GC, and MS-based analysis of synthetic products

## Teaching and leadership

2020–Present **Live-in Resident Tutor**, *New Vassar House, MIT*, Cambridge, MA

- Helped guide and support a community of $\sim 40$ undergraduate students
- Provided academic advising, address concerns among students, facilitate a safe living community

2018–Present **Statistics and Medical School Admissions Tutor**, *Cambridge Coaching*, Cambridge, MA

2021, 2022 **Journal Club Facilitator**, *The YES for CURE Program*, Cambridge, MA

- Ran a weekly cancer-focused journal club for high schoolers emphasizing paper reading skills

2017–2021 **Class Representative to the Curriculum Committee**, *HST MD Program, Harvard Medical School*, Boston, MA

2018–2020 **Non-resident Pre-medical Tutor**, *Kirkland House, Harvard University*, Cambridge, MA

2018–2019 **Co-founder, MD HST Diversity Ambassadors**, *HST MD Program, Harvard Medical School*, Boston, MA

- Identified steps in the HST application pipeline which exacerbated exclusion of minority groups
- Designed targeted materials to boost engagement with underrepresented schools and groups

2014–2015 **Teaching assistant**, *Introduction to Engineering, UPenn SEAS*, Philadelphia, PA

2015 **Teaching assistant**, *Mass Balances, UPenn SEAS*, Philadelphia, PA

2014–2016 **Founder and co-president of Access Engineering**, *UPenn SEAS*, Philadelphia, PA

- Developed the organization's vision to improve engineering outreach to underprivileged high schools through weekly hands-on labs
- Created a 2-semester curriculum including Java, Arduino robotics, and computer-aided design
- Formed relationships with 5 high schools, recruiting >70 students for weekly lessons
- Won School of Engineering and Applied Sciences club of the year in 2016

2014–2016 **New Initiatives Chair and Section Leader of Penn Sciences Across Ages**, *University of Pennsylvania*, Philadelphia, PA

- Hosted a weekly science class at Lea Day School
- Worked on expanding outreach efforts by contacting new schools and developing new programs

2014–2016 **Community Service Chair**, *UPenn Tau Beta Pi Engineering Honor Society*, Philadelphia, PA

## Skills

Graduate coursework: Probability I (BST 230), Statistical Inference I (BST 231), Statistical Methods (BST 232), Bayesian Methodology (BST 249), Principles of Genetics (GEN 201)

Programming: R, Python, MATLAB, Java, OCaml, M/Caché

Large scale workflows: Biobank-scale genome-wide association studies, multi-ancestry heritability analyses, phenome-wide association analyses, cross-trait genotype and phenotype correlations

Platforms: Google Cloud Platform, DNANexus, Amazon AWS, Terra

Tools: ImageJ, SolidWorks, ImageStudio, Adobe Illustrator, Docker