

## 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)  
**Majors:** Chemical & Biomolecular Engineering, Biology; **Minor:** Chemistry

## 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 Therapeutics**

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

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## 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*

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## 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

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## 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  $\approx 10,000$  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 **Cytect 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 ~ 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