

Yusuph Mavura, PhD, MS

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EDUCATION

- Sep 2019 – Mar 2025 **Doctor of Philosophy in Epidemiology and Translational Science**
San Francisco, CA *University of California, San Francisco*
- Aug 2017 - Dec 2018 **Master of Science in Bioinformatics**
Atlanta, GA *Georgia Institute of Technology*
- May 2012 - May 2016 **Bachelor of Science in Biochemistry and Molecular Biology**
Nairobi, Kenya *Jomo Kenyatta University of Agriculture and Technology*
• Summa Cum Laude

RESEARCH EXPERIENCE

- Jan 2024 – Mar 2025 **NIH F99/K00 Fellow & Dissertation Student** / University of California, San Francisco
Project Advisors: Drs. Neil Risch, Maria Glymour, Akinyemi Oni-Orisan, Catherine Tcheandjie, Franklin Huang
- Leveraged longitudinal real-world electronic health records (EHR) data to assess the clinical utility of multi-ancestry polygenic scores
 - Designed and implemented pipelines integrating Kaiser Permanente genotype and EHR data using R/Python/Bash, and high-performance computing (HPC)
 - Led and designed analyses of environmental, and genetic, effects on antihypertensive monotherapy response in All of Us and Kaiser Permanente data
 - Authored three dissertation manuscripts and maintained all analysis code in GitHub for reproducibility, version control and collaboration
- Mar 2020 – Jan 2023 **Graduate Student Rotation** / University of California, San Francisco
Project Advisors: Dr. John Witte, Dr. Franklin Huang
- Investigated pleiotropy in a pan-cancer, cross-tissue transcriptome-wide association study (TWAS) using UKBiobank and Kaiser Permanente's cohort
 - Led a research project characterizing the genomic rearrangement landscape in localized prostate cancer samples from Nigerian men
 - First author manuscript published in *Prostate*, collaborating with researchers in Nigeria
- Sep 2019 – Mar 2025 **Graduate Student Researcher** / University of California, San Francisco
Project Advisor: Dr. Neil Risch
- Assessed diagnostic yield of exome and genome sequencing, finding no evidence of differences by genetic ancestry in cases with suspected mendelian disorders.
 - Processed and cleaned genetic, clinical, survey data from the Clinical Sequencing Evidence-Generating Research (CSER) Phase II consortium for analysis
 - Designed and implemented genetic ancestry, relatedness, and consanguinity estimation analyses for multiple nationwide consortia studies
 - Collaborated and coordinated with interdisciplinary teams of consortia leadership, clinicians, database administrators, ethics boards nationwide and shared findings
 - First authored and co-authored manuscripts published in *npj Genomics*; with multiple other manuscripts in review.
- Jun 2018 – Aug 2018 **Intern** / Department of Cancer Epidemiology and Genetics (DCEG), National Cancer Institute (NCI), NIH
Project Advisor: Dr. Laufey Amundadottir
- Conducted a Genome-Wide Association study (GWAS) which identified multiple new risk loci for pancreatic cancer in East Asian populations
 - Developed and executed data pipelines in PLINK and R for population-stratification checks and covariate adjustment

- Generated GWAS summary statistics and visualizations, including Manhattan, Q–Q, and regional association plots
- Drafted methods and results sections for internal reports, NIH progress updates, and manuscript submissions

Aug 2017 – **Graduate Student Researcher** / Georgia Institute of Technology
Dec 2018 *Project Advisor: Dr. John McDonald*

- Performed variant calling and annotation on matched primary, metastatic, and recurrent ovarian tumor Exome Sequencing samples using GATK best practices
- Recovered, extracted, and maintained sequencing and clinical data on the lab's secure local server
- Curated a catalogue of known and candidate cancer driver genes (e.g., TP53, BRCA1/2, NF1) for downstream analyses
- Presented results in lab meetings, preparing slides and detailed reports to communicate findings to supervisors and colleagues

PUBLICATIONS

- **Yusuph Mavura**, Nuriye Sahin-Hodoglugil, Ugur Hodoglugil, Mark Kvale, Pierre-Marie Martin, Jessica Van Ziffle, W. Patrick Devine, Sara L. Ackerman, Barbara A Koenig, Pui-Yan Kwok, Mary E. Norton, Anne Slavotinek, Neil Risch. Genetic ancestry and diagnostic yield of exome sequencing in a diverse population. *npj Genom. Med.* 9, 1 (2024). <https://doi.org/10.1038/s41525-023-00385-6>.
- Slavotinek A, Rego S, Sahin-Hodoglugil N, Kvale M, Lianoglou B, Yip T, Hoban H, Outram S, Anguiano B, Chen F, Michelson J, Cilio RM, Curry C, Gallaher RC, Gardener M, Kuperman R, Mendelsohn B, Sherr E, Shieh J, Strober J, Tam A, Tenney J, Weiss W, Whittle A, Chin G, Faubel A, Prasad H, **Mavura Y**, Van Ziffle J, Devine WP, Hodoglugil U, Martin PM, Sparks TN, Koenig B, Ackerman S, Risch N, Kwok PY, Norton ME. Diagnostic yield of pediatric and prenatal exome sequencing in a diverse population. *NPJ Genom. Med.* 2023 May 26;8(1):10.doi: 10.1038/s41525-023-00353-0.PubMed PMID: 37236975; PubMed Central PMCID: PMC10220040.
- **Mavura Y**, Song H, Xie J, Tamayo P, Mohammed A, Lawal AT, Bello A, Ibrahim S, Faruk M, Huang FW. Transcriptomic profiling and genomic rearrangement landscape of Nigerian prostate cancer. *Prostate.* 2023 Jan 4; doi: 10.1002/pros.24471. PubMed PMID: 36598071.
- Kamran SC, Xie J, Cheung ATM, **Mavura MY**, Song H, Palapattu EL, Madej J, Gusev A, Van Allen EM, Huang FW. Tumor Mutations Across Racial Groups in a Real-World Data Registry. *JCO Precis Oncol.* 2021 Nov;5:1654-1658. doi: 10.1200/PO.21.00340. PMID: 34994651.
- Oni-Orisan A, **Mavura Y**, Banda Y, Thornton TA, Sebro R. Embracing Genetic Diversity to Improve Black Health. *N Engl J Med.* 2021 Mar 25;384(12):1163-1167. doi: 10.1056/NEJMs2031080. Epub 2021 Feb 10. PMID: 33567186.
- **Mavura MY**, Huang FW. How Cancer Risk SNPs May Contribute to Prostate Cancer Disparities. *Cancer Res.* 2021 Jul 15;81(14):3764-3765. doi: 10.1158/0008-5472.CAN-21-1146. PMID: 34266915.

Publications Under Review

- **Yusuph Mavura**, David Crosslin, Kathleen DM Ferar, John Greally, Lucia Hindorff, Gail P Jarvik, Sara Kalla, Barbara A Koenig, Mark Kvale, Pui-Yan Kwok, Mary Norton, Sharon E. Plon, Bradford C. Powell, Anne Slavotinek, Michelle L Thompson, Alice B Popejoy, Eimear E. Kenny, Neil Risch. *Diagnostic Yield Results from the CSER 2 consortium: no evidence for differences by genetic ancestry.* (Under Review at *PLOS Genetics*).
- Beale, H. C., Tse, V., Lee, J. Y., Akutagawa, J., **Mavura, Y.**, Saint-John, B., Cheney, A., Mulligan, D. R., Chacaltana, G., Gutierrez, M., Tenney, J., Shieh, J. T., Martin, P.-M., Yip, T., Hodoglugil, U., Fay, A. J., Brooks, A. N., Ziffle, J. V., Stone, M. D., ... Slavotinek, A. (2025). *A novel splice site variant in DEGS1 leads to aberrant splicing and loss of DEGS1 enzyme activity, a VUS resolved* (p. 2025.04.04.25325118). medRxiv. <https://doi.org/10.1101/2025.04.04.25325118>. Under Review.

SELECTED CONFERENCE ABSTRACTS

- **Yusuph Mavura**, David Crosslin, Kathleen DM Ferar, John Grealley, Lucia Hindorff, Gail P Jarvik, Sara Kalla, Barbara A Koenig, Mark Kvale, Pui-Yan Kwok, Mary Norton, Sharon E. Plon, Bradford C. Powell, Anne Slavotinek, Michelle L Thompson, Alice B Popejoy, Eimear E. Kenny, Neil Risch. *Diagnostic yield of genome and exome sequencing in the ancestrally diverse CSER Phase II consortium is not associated with genetic ancestry in a variety of clinical settings*. American Society of Human Genetics, Los Angeles, CA. October 2023.
- **Yusuph Mavura**, Nuriye Sahin-Hodoglugil, Mark Kvale, Jessica Van Ziffle, W. Patrick Devine, Ugur Hodoglugil, Pierre-Marie Martin, Barbara A Koenig, Sara Ackerman, Anne Slavotinek, Pui-Yan Kwok, Mary E. Norton, Neil Risch. *No reduction in diagnostic yield of exome sequencing in prenatal and pediatric patients with non-European ancestries*. American Society of Human Genetics, Los Angeles, CA. October 2022.

RESEARCH SUPPORT

1F99HG013437-01 Assessing Clinical Utility of Polygenic Risk Scores in Ancestrally Diverse Real-World Cohorts
Mavura (PI)
Sponsor and Co-Sponsors: Drs. Neil Risch, Maria Glymour, Akinyemi Oni-Orisan, Catherine Tcheandjieu

AWARDS AND HONORS

2024 NIH F99/K00 Predoctoral to Postdoctoral Transition Award Fellow (Highest possible impact score of 10)
2023 NIH T32 Aging for Research on Aging and Chronic Disease (Trainee), UCSF
2018 Computational Biology Faculty GRA Award, *Georgia Institute of Technology*

ORAL PRESENTATIONS

Jan 2025 Invited Guest Speaker, NIH NHGRI Childhood Complex Disease Genomics Section, Hanchard Lab: "Blood pressure polygenic scores, and antihypertensive drug effects: Insights from real world longitudinal data"
Nov 2024 PhD dissertation defense seminar, UCSF "Blood pressure polygenic scores, and antihypertensive drug effects: Insights from real world longitudinal data"
Jun 2021 NIH CSER Spring 2021 virtual meeting Junior Investigator Presentations, "Determining Genetic Ancestry of P³EGS Participants using Exome-Wide Markers"
Nov 2015 Undergraduate thesis defense, "*In silico* validation of mutations in APIAP2 gene and structural consequence of the mutations in piperazine and lumefantrine resistant *plasmodium berghei* anka"

TEACHING AND MENTORSHIP EXPERIENCE

University of California, San Francisco (SF, California)

Mar – Jun 2022 **Teaching Assistant;** Advanced Approaches to Analysis of Observational Data
Jan – Mar 2021 **Teaching Assistant;** Molecular and Genetic Epidemiology

PROFESSIONAL MEMBERSHIPS

2020- American Society of Human Genetics (ASHG)
2025- American Society of Human Genetics (ASHG) Career Development Committee member