

STEPHANIE M. YAN

Computational biologist specializing in human genomics & evolution

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SUMMARY

- Computational geneticist with 5+ years of experience in human genomics, data science, and analysis of large genomic & transcriptomic datasets.
- Fast and adaptable learner who began graduate school with no genetics or computational background and rapidly grew into a skilled researcher and programmer.
- Excellent scientific writer and communicator, with ten publications and five talks at national and international conferences during graduate school.

EDUCATION

Johns Hopkins University

Ph.D. in Cell, Molecular, Developmental Biology and Biophysics

2018 – 2024

Baltimore, MD

Cornell University

B.A. in Biological Sciences with Distinction, *magna cum laude*

2014 – 2018

Ithaca, NY

TECHNICAL SKILLS

Genomics: Variant calling & genotyping, long- and short-read genomics & transcriptomics, e/sQTL mapping

Population genetics: Evolutionary simulation, scans for selection & introgression, archaic hominin genomics

Scientific writing: Ten publications (four as first or co-first author), three awarded grants

Scientific communication: Six talks at international conferences & symposia

Programming: R, Bash, Python, high-performance computing, SLiM, Git, Java

AWARDS & HONORS

- **NRSA (F31) Graduate Fellowship;** NIH/NHGRI
- **Honorable Mention;** National Science Foundation Graduate Research Fellowships Program (NSF GRFP)
- **Adam T. Bruce Fellowship;** Johns Hopkins University
- **Registration Award;** Society for Molecular Biology & Evolution

RESEARCH EXPERIENCE

Novel RNA isoform discovery from long-read sequencing | Johns Hopkins University

2022 – pres.

- Leading a three-lab collaboration at Johns Hopkins to generate and analyze paired long-read transcriptomics and proteomics data from geographically diverse human samples.
- Co-led a successful \$100,000 JHU Discovery Grant application for this project.
- Research was featured by Oxford Nanopore Technologies in an interview at the ASHG 2023 conference.

Evolutionary simulations of somatic mosaicism | Johns Hopkins University

2022 – pres.

- Developed the first model of somatic evolution in humans that incorporates telomere length as a senescence factor.
- Demonstrated that telomere length alone increases the risk of accumulating harmful somatic mutations and cancer.
- Published work in *NEJM*, in collaboration with researchers at the Johns Hopkins School of Medicine.

Telomere-to-Telomere (T2T) Consortium | Johns Hopkins University

2021 – 2022

- Led investigation of variants in novel regions of the T2T reference genome, requiring extensive analysis of variants in complex centromeric, telomeric, and repetitive sequences.
- Co-first authorship on the T2T variant calling paper, as well as authorship on the T2T flagship paper, both of which were published in *Science*.

Local adaptation at human structural variant loci | Johns Hopkins University

2019 – 2021

- Combined long-read sequencing and variant graph genotyping to discover structural variants in 2,504 individuals from the 1000 Genomes Project.
- Identified recent and strong selection on a Neanderthal-introgressed haplotype in southeast Asian populations.
- Results were published in an *eLife* Special Issue and selected for presentations at multiple international conferences.

PUBLICATIONS & PRESENTATIONS

SELECTED PUBLICATIONS

- Taylor DJ, Chhetri SB, Tassia MG, Biddanda A, **Yan SM**, *et al.* (2024). Sources of gene expression variation in a globally diverse human cohort. *Nature*, in press.
- DeBoy EA, Tassia MG, Schratz KE, **Yan SM**, *et al.* (2023). Familial Clonal Hematopoiesis in a Long Telomere Syndrome. *NEJM*, 388: 2422–2433. DOI: 10.1056/NEJMoa2300503.
- Aganezov S*, **Yan SM***, Soto DC*, Kirsche M*, Zarate S*, *et al.* (2021). A complete reference genome improves analysis of human genetic variation. *Science*, 376(6588): 54. DOI: 10.1126/science.abl3533. *Co-first author
- Nurk S, Koren S, Rhie A, Rautiainen M, ... , **Yan SM**, *et al.* (2021). The complete sequence of a human genome. *Science*, 376(6588): 44–53. DOI: 10.1126/science.abj6987.
- Yan SM**, Sherman RM, Taylor DJ, Nair DR, Bortvin AN, Schatz MC, & McCoy RC. (2021). Local adaptation and archaic introgression shape global diversity at human structural variant loci. *eLife*, 10: e67615. DOI: 10.7554/eLife.67615.
- Yan SM** & McCoy RC. (2020). Archaic hominin genomics provides a window into gene expression evolution. *Curr. Opin. Genet. Dev.*, 62: 44–49. DOI: 10.1016/j.gde.2020.05.014.

SELECTED ORAL PRESENTATIONS

- 2022 Advances in Genome Biology and Technology
- 2021 eLife Symposium: Evolutionary Medicine
- 2021 Cold Spring Harbor Laboratory: Biology of Genomes
- 2020 American Society of Human Genetics
- 2020 Society for Molecular Biology & Evolution | *Cancelled due to COVID-19*

SCIENTIFIC COMMUNICATION

Technology Fellow | *Johns Hopkins University Center for Teaching Excellence and Innovation* 2021 – 2023

- Awarded a \$4,000 grant to develop open-source teaching materials in R, intended to introduce undergraduates with no coding experience to human genetics research.
- Created modules for a semester-long course on genomic data analysis, as well as an accompanying digital textbook.
- Published all materials on Github as a community resource: github.com/mccoy-lab/hgv_modules.

Teaching Assistant | *Quantitative Biology Bootcamp & Lab, Johns Hopkins University* 2020 – 2022

- Teaching assistant for the Department of Biology's introductory bioinformatics courses for first-year PhD students, taught in Python and bash.
- Worked with other TAs to adapt these coding-intensive courses, normally dependent on in-person assistance, into remote and hybrid formats during the pandemic.
- Consistently highlighted in anonymous student evaluations as an integral part of the course's success.

Assistant News Editor | *The Cornell Daily Sun* 2014 – 2018

- Collaborated in a six-person news editor team to publish the paper's daily news section, cover breaking news, develop investigative pieces, and train writers.
- Led the transition to web-first reporting and social media during the paper's shift from print to web publication.

LEADERSHIP & SERVICE

BioRep | *Department of Biology, Johns Hopkins University* 2018 – 2024

- Elected to represent my graduate cohort (17 students) and advocate for them to the program directors and administrators of the biology PhD program.
- Interfaced with JHU and program leadership to address department climate issues and student concerns.

Co-Founder & Accountability Leader | *symBIOsis, Johns Hopkins University* 2020 – 2023

- Co-founder of symBIOsis, a student organization dedicated to the mentorship and support of 120+ graduate students in the Department of Biology.
- Served as the symBIOsis Accountability Leader for two years, managing the organization's committee members, events, and communications with JHU administration.