

# STEPHANIE M. YAN

Computational biologist specializing in human genomics & evolution

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## SUMMARY

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- Computational geneticist with 5+ years of experience in human genomics, evolutionary biology, 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, three awarded grants, and six talks at national and international conferences during graduate school.

## EDUCATION

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### 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

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**Genomics:** SNV/indel and structural variant calling & genotyping, analysis of long- & short-read sequencing data

**Transcriptomics:** e/sQTL mapping, fine mapping, novel isoform discovery from long reads

**Population genetics:** Evolutionary simulation, scans for selection & introgression, local ancestry

**Programming:** R, Bash, Python, high-performance computing, Git, AnVIL/Terra, Java

**Bioinformatics software:** bcftools, plink, GATK, bedtools, IGV, SLiM, fastQTL, StringTie, FLAIR

## AWARDS & HONORS

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- **Stephen and Carolyn Oppenheimer Thesis Award;** Johns Hopkins University Department of Biology
- **NRSA (F31) Graduate Fellowship;** NIH/NHGRI
- **Honorable Mention;** National Science Foundation Graduate Research Fellowships Program (NSF GRFP)
- **Adam T. Bruce Fellowship;** Johns Hopkins University

## RESEARCH EXPERIENCE

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### 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

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### 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 TALKS

- 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

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**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](https://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

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**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.