Curriculum Vitae

Stephanie M. Yan

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Education

2018–2024 **Johns Hopkins University**, Baltimore, MD
PhD Program in Cell, Molecular, Developmental Biology, and Biophysics (CMDB)

2014–2018 **Cornell University**, Ithaca, NY
B.A. in Biological Sciences with Distinction in all Subjects; Linguistics Minor
Honors Thesis in Molecular & Cell Biology, *magna cum laude*
GPA: 3.84 / 4.00

Research experience

2024–pres. **Postdoctoral Fellow**, Johns Hopkins University, Baltimore, MD
Advisor: Rajiv McCoy

- Leading a three-lab collaboration at Johns Hopkins to generate and analyze paired 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.

2018–2024 **NRSA (F31) Predoctoral Fellow**, Johns Hopkins University, Baltimore, MD
Advisor: Rajiv McCoy

- Leveraged short- and long-read sequencing data to characterize the functional impacts of genetic variants in large genomic and transcriptomic datasets.
- Developed a WGS analysis pipeline, combining long-read sequencing and variant graph genotyping, to discover structural variants in a dataset of 2,504 individuals.
- Extensive experience in transcriptomics (e/sQTL mapping, fine mapping, effect size estimation), including work currently in press at *Nature*.
- Collaborated with the Telomere-to-Telomere Consortium to lead functional characterization of novel variants in the T2T reference genome.
- 10 publications and 6 talks at national and international conferences.

2016–2018 **Undergraduate Researcher**, Cornell University, Ithaca, NY
Advisor: Scott Emr

- Studied the regulation of transmembrane nutrient transporters by arrestin-family proteins in yeast.

- As part of my honors thesis research, I carried out and co-designed a screen for identifying arrestin binding sites.
- This method was included in a Methods in Enzymology book chapter and taught in a lab course at Hartwick College.

2015 **Research Trainee**, H. Lee Moffitt Cancer Center & Research Institute, Tampa, FL
Advisor: Shengyu Yang
- Worked with cancer cell lines to study the impact of deoxyguanosine kinase (DGUOK), a mitochondrial protein, on the regulation of cancer metabolism and metastasis.

Publications

Research and review articles (peer reviewed)

2024 Taylor, D. J., Chhetri, S. B., Tassia, M. G., Biddanda, A. A., **Yan, S. M.**, Wojcik, G. L., Battle, A. J., & McCoy, R. C. (2024). Sources of gene expression variation in a globally diverse human cohort. *Nature*, in press.

2023 DeGorter, M. K.*, Goddard, P. C.*, Karakoc, E., Kundu, E., Kundu, S., **Yan, S. M.**, ... , & Montgomery, S. B. (2023). Transcriptomics and chromatin accessibility in multiple African population samples. *bioRxiv*, DOI: 10.1101/2023.11.04.564839.

2023 DeBoy, E. A.*, Tassia, M. G.*, Schratz, K. E., **Yan, S. M.**, Cosner, Z. L., McNally, E. J., Gable, D. L., Xiang, Z., Lombard, D. B., Antonarakis, E. S., Gocke, C. D., McCoy, R. C., & Armanios, M. (2023). Familial Clonal Hematopoiesis in a Long Telomere Syndrome. *NEJM*, 388:2422-2433. DOI: 10.1056/NEJMoa2300503.

2022 Aganezov, S.*, **Yan, S. M.***, Soto, D. C.*, Kirsche, M.*, Zarate, S.*, Avdeyev, P., Taylor, D. J., Shafin, K., Shumate, A., Xiao, C., Wagner, J., McDaniel, J., Olson, N. D., Sauria, M. E. G., Vollger, M. R., Rhie, A., Meredith, M., Martin, S., Koren, S., Rosenfeld, J. A., Paten, B., Layer, R., Chin, C., Sedlazeck, F. J., Hansen, N. F., Miller, D. E., Phillippy, A. M., Miga, K. H., & McCoy, R. C.*, Dennis, M. Y.*, Zook, J. M.*, Schatz, M. C.* (2022). A complete reference genome improves analysis of human genetic variation. *Science*, *376*(6588): 54, DOI: 10.1126/science.abl3533.

2022 Nurk, S.*, Koren, S.*, Rhie, A.*, Rautiainen, M.*, ... , **Yan, S. M.**, ... , & Eichler, E. E.*, Miga, K. H.*, Phillippy, A. M.* (2022). The complete sequence of a human genome. *Science*, *376*(6588): 44–53, DOI: 10.1126/science.abj6987.

2021 Ariad, D., **Yan, S. M.**, Victor, A. R., Barnes, F. L., Zouves, C. G., Viotti, M., & McCoy, R. C. (2021). Haplotype-aware inference of human chromosome abnormalities. *Proceedings of the National Academy of Sciences*, *118*(46): e2109307118. DOI: 10.1073/pnas.2109307118

2021 **Yan, S. M.**, Sherman, R. M., Taylor, D. J., Nair, D. R., Bortvin, A. N., Schatz, M. C., & McCoy, R. C. (2021). Local adaptation and archaic introgression shape global diversity at human structural variant loci. *eLife*, *10*: e67615. DOI: 10.7554/eLife.67615.

2020 **Yan, S. M.** & McCoy, R. C. (2020). Archaic hominin genomics provides a window into gene expression evolution. *Current Opinion in Genetics & Development, 62*: 44–49. DOI: 10.1016/j.gde.2020.05.014.

Editorials and commentaries

2024 Taylor, D. J.* & **Yan, S. M.*** (2024). Diversifying human gene expression research. *Nature (Research Briefing)*, in press.

2023 Soto, D.C.*, Kirsche, M.K.*, **Yan, S. M.***, Zarate, S.*. (2022), The human reference genome is finally complete. *The Science Breaker*. DOI: 10.25250/thescbr.brk721.

2019 **Yan, S. M.** & McCoy, R. C. (2019), Functional divergence among hominins. *Nature Ecology & Evolution (News & Views), 3*: 1507–1508. DOI: 10.1038/s41559-019-0995-y.

**Equal contribution*

Presentations

Oral presentations

2022 **JHU CMDB Program Retreat**, Harpers Ferry, VA (invited talk)
Local adaptation and archaic introgression at human structural variant loci

2022 **T2T-F2F**, Santa Cruz, CA (flash talk)
Human genetic diversity within challenging regions of the genome

2022 **Advances in Genome Biology and Technology**, Orlando, FL (selected abstract)
A complete reference genome improves analysis of human genetic variation

2021 **eLife Symposium: Evolutionary Medicine** (virtual)
Local adaptation and archaic introgression at human structural variant loci

2021 **Cold Spring Harbor Laboratory: Biology of Genomes** (virtual)
Local adaptation and archaic introgression at human structural variant loci

2020 **American Society of Human Genetics** (virtual)
The role of structural variation in human local adaptation

*2020 **Society for Molecular Biology & Evolution**, Québec City, Canada
The role of structural variation in human local adaptation

**Cancelled due to COVID-19*

Poster presentations

2024 **Cold Spring Harbor Laboratory: Biology of Genomes**, Cold Spring Harbor, NY
A high-resolution view of human gene expression and splicing diversity with long-read sequencing

2023 **American Society of Human Genetics**, Washington D.C.
A high-resolution view of human gene expression and splicing diversity with long-read sequencing

2023 **Society for Molecular Biology & Evolution**, Ferrara, Italy (virtual)
Evolutionary simulations inform the origins of clonal hematopoiesis

2020 **The Allied Genetics Conference** (virtual)
The role of structural variation in human local adaptation

2019 **13th Annual Genomics and Bioinformatics Symposium**, Baltimore, MD
Structural genomic divergence and introgression in hominin evolution

2019 **Society for Molecular Biology & Evolution**, Manchester, U.K.
Structural genomic divergence and introgression in hominin evolution

2018 **Biological Sciences Honors Symposium**, Ithaca, NY
Regulation of plasma membrane proteins by the Art1-Rsp5 ubiquitin ligase complex

Grants

2022–2024 **JHU Discovery Award**: JHU grants for cross-divisional collaborations
PIs: Rajiv McCoy, Winston Timp, Alexis Battle
Title: "A high-resolution view of human gene expression and splicing diversity with single-molecule long- read sequencing."

2022–2024 **NIH/NHGRI F31**: Ruth L. Kirschstein Predoctoral National Research Service Award
PI: Stephanie Yan
Title: "Investigating the role of structural variation in hominin evolution."

2021–2023 **Technology Fellowship Grant**: JHU Center for Teaching Excellence and Innovation
PIs: Rajiv McCoy, Stephanie Yan, Kate Weaver
Title: "Cloud-based modules for interactive exploration of human genomic variation."

Fellowships, honors, & awards

2024 Stephen and Carolyn Oppenheimer Thesis Award; JHU Department of Biology

2023 Margolies Travel Award; JHU CMDB Program

2022 Ruth L. Kirschstein National Research Service Award (NRSA) Fellowship; NHGRI

2020 Technology Fellowship Grant; JHU Center for Educational Resources

2020 Travel Grant; JHU Graduate Representative Organization

2020 Honorable Mention; NSF Graduate Research Fellowships Program

2019 Adam T. Bruce Fellowship; Johns Hopkins University

2019 Registration Award; Society for Molecular Biology & Evolution

2014–2018 Tanner Dean's Scholar; Cornell University

2017 Book Prize for Neuropathology; St Catherine's College, University of Oxford (study abroad)

Teaching

2024 Teaching Assistant, Biology REU Data Science Workshop, Johns Hopkins University

2019-2024 Guest Lecturer, Human Genome Variation Lab, Johns Hopkins University

2021–2023 Tutor, Quantitative Biology Lab, Johns Hopkins University

2023 Guest Speaker, Population Genetics Simulation and Visualization, Johns Hopkins University

2021–2023 Technology Fellow, Human Genome Variation Lab, Johns Hopkins University

2020–2022 Teaching Assistant, Quantitative Biology Bootcamp, Johns Hopkins University

2022 Guest Lecturer, Human Genome Variation, Johns Hopkins University

2020 Teaching Assistant, Quantitative Biology Lab, Johns Hopkins University

2019–2020 Teaching Assistant, Developmental Genetics Lab, Johns Hopkins University

Research mentoring

PhD Rotation Students

2023 Betty Huang, JHU CMDB program

2023 Catherine Brown, JHU CMDB program

2023 Emily Shen, JHU CMDB program

2022 J. Noah Workman, JHU SOM Human Genetics program

2019 Sara Carioscia, JHU CMDB program

Undergraduate

2020–2023 Divya Nair, Johns Hopkins University

High school

2020–2021 Miles Fancher, Ingenuity Project, Baltimore Polytechnic Institute

2020–2021 Aram Zaprosyan, Ingenuity Project, Baltimore Polytechnic Institute

Academic service

Academic service

2018–2024 **BioRep**, CMDB Program, Johns Hopkins University
- 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 and led initiatives to address department climate issues and student concerns.

2023 **CMDB Representative**, NIH Graduate & Professional School Fair, Bethesda, MD

2020–2023 **Co-Founder and Accountability Leader**, CMDB symBIOsis, Johns Hopkins University
- Co-founder of symBIOsis, a student organization dedicated to the mentorship and support of 120+ graduate students in the Department of Biology.
- Responsible for organizing the BioBuddies peer mentorship program, advising meetings on program requirements, and social events for CMDB graduate students.
- Served as the symBIOsis Accountability Leader for two years, managing the organization's committee members, events, and communications with JHU administration.

2021 **Session Moderator**, American Society of Human Genetics (virtual)
- Session: Introgression and population structure in the age of genomic biobanks

2020 **James Taylor Memoriam Committee**, Johns Hopkins University
- Led the creation and maintenance of jxtx-memorial.github.io, a memorial website for Prof. James Taylor in the Department of Biology.

2019 **Session Moderator**, Johns Hopkins CMDB Program Retreat, Fairfield, PA

Peer mentorship

2019–2023 **BioBuddies Mentor**, CMDB symBIOsis, Johns Hopkins University

2019–2020 **BioBuddies Program Co-Chair**, CMDB MInDS, Johns Hopkins University

2019 **Biology REU Mentor**, CMDB MInDS, Johns Hopkins University

2016–2017 **College of Arts & Sciences Peer Advisor**, Cornell University

Volunteer work & outreach

2018–2023 **2018 Class Correspondent**, Cornell Association of Class Officers

- Developed communications and events, including a quarterly newsletter, Instagram account, and quarterly updates in the Cornell Alumni Magazine, to maintain engagement with alumni from the Class of 2018.

2019–2020 **Webmaster & Social Media Chair**, CMDB MInDS, Johns Hopkins University

- Maintained website and social media accounts for Mentoring to Inspire Diversity in Science (MInDS), a group that organizes community outreach and student support activities for members of the biology department.
- Helped lead the 2020 restructuring of MInDS into two groups, one (MInDS) focusing on outreach and diversity and the other (symBIOsis) focusing on student support.

Technical writing & editing

2015–2018 **Assistant News Editor, Print Designer, & Staff Writer**, *The Cornell Daily Sun*, Ithaca, NY

- Wrote for, edited, and designed The Sun, an independent, student-run paper and the primary source of news on the Cornell campus.
- Collaborated in a six-person news editor team to publish the paper's daily news section, cover breaking news, assign long-term investigative pieces, train writers, and hold semiweekly workshops.

2015–2018 **Managing Editor, Copy Editor, & Staff Writer**, *The Research Paper*, Ithaca, NY

- Wrote for and edited a student-run magazine on undergraduate research at Cornell.
- Recruited researchers to be featured; trained new writers; and corrected articles for grammar, clarity, and technical accuracy.

2017 **Staff Writer**, *Bang! Science Magazine*, Oxford, U.K.

- Wrote feature pieces explaining newly published research to a non-scientist readership.

2015 **Editorial Intern**, *Familius*, Sanger, CA

- Worked with authors to edit nonfiction manuscripts at the substantive editing, copy editing, and proofreading stages.
- Credited as the primary editor on three Familius titles.