

Mitchell R. Vollger

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Postdoctoral Scholar in the Division of Medical Genetics at the University of Washington

Education

Ph.D. in Genome Sciences at University of Washington

Sep 2016 - March 2021

Dissertation: Assembly of segmental duplications and their variation in humans

Seattle, Washington

- Advisor: Evan E. Eichler
- Completed the [Advanced Data Science Option](#)

B.S.E. in Computer Science Engineering at Princeton University

Sep. 2011 - June 2015

Departments of Computer Science and Quantitative and Computational Biology

Princeton, New Jersey

- Student of the [Integrated Science Curriculum](#)
- Certificate in Quantitative and Computational Biology

Associate of Arts Degrees at College of the Redwoods

Sep. 2008 - June 2011

AA in Mathematics | AA in Science

Eureka, California

Postdoctoral Experience

Postdoctoral Scholar in the Division of Medical Genetics

April 2022 - Present

In the lab of Andrew B. Stergachis

University of Washington

Postdoctoral Scholar in the Department of Genome Sciences

March 2021 - April 2022

In the lab of Evan E. Eichler

University of Washington

Funding and Awards

K99/R00 Pathway to Independence Award

Summer 2024 - present

National Institute of General Medical Sciences, 1K99GM155552-01

Seattle, Washington

NIH/NHGRI T32 Genome Training Grant

Fall 2022 - Fall 2024

Division of Medical Genetics at University of Washington

Seattle, Washington

BDGN, Big Data in Genomics and Neuroscience

Fall 2017 - Fall 2019

Genome Sciences at University of Washington

Seattle, Washington

NIH/NHGRI T32 Genome Training Grant

Fall 2016 - Fall 2017

Genome Sciences at University of Washington

Seattle, Washington

Publications

- First Author** [M. R. Vollger](#), E. G. Swanson, S. J. Neph, J. Ranchalis, K. M. Munson, C.-H. Ho, A. E. Sedeño-Cortés, W. E. Fondrie, S. C. Bohaczuk, Y. Mao, N. L. Parmalee, B. J. Mallory, W. T. Harvey, Y. Kwon, G. H. Garcia, K. Hoekzema, J. G. Meyer, M. Cicek, E. E. Eichler, ... A. B. Stergachis, A haplotype-resolved view of human gene regulation (2024), doi: [10.1101/2024.06.14.599122](https://doi.org/10.1101/2024.06.14.599122)
- [M. R. Vollger](#), J. Korlach, K. C. Eldred, E. Swanson, J. G. Underwood, Y.-H. H. Cheng, J. Ranchalis, Y. Mao, E. E. Blue, U. Schwarze, K. M. Munson, C. T. Saunders, A. M. Wenger, A. Allworth, S. Chanprasert, B. L. Duerden, I. Glass, M. Horike-Pyne, M. Kim, ... A. B. Stergachis, Synchronized long-read genome, methylome, epigenome, and transcriptome for resolving a Mendelian condition. *Nature Genetics*, *accepted in principle* (2024), doi: [10.1101/2023.09.26.559521](https://doi.org/10.1101/2023.09.26.559521)
- [M. R. Vollger](#), P. C. Dishuck, W. T. Harvey, W. S. DeWitt, X. Guitart, M. E. Goldberg, A. N. Rozanski, J. Lucas, M. Asri, H. P. R. Consortium, K. M. Munson, A. P. Lewis, K. Hoekzema, G. A. Logsdon, D. Porubsky, B. Paten, K. Harris, P. Hsieh, E. E. Eichler, Increased mutation and gene conversion within human segmental duplications. *Nature*. **617**, 325–334 (2023)
- [M. R. Vollger](#), X. Guitart, P. C. Dishuck, L. Mercuri, W. T. Harvey, A. Gershman, M. Diekhans, A. Sulovari, K. M. Munson, A. P. Lewis, K. Hoekzema, D. Porubsky, R. Li, S. Nurk, S. Koren, K. H. Miga, A. M. Phillippy, W. Timp, M. Ventura, E. E. Eichler, Segmental duplications and their variation in a complete human genome. *Science*. **376** (2022), doi: [10.1126/science.abj6965](https://doi.org/10.1126/science.abj6965)
- [M. R. Vollger](#), P. Kerpedjiev, A. M. Phillippy, E. E. Eichler, StainedGlass: Interactive visualization of massive tandem repeat structures with identity heatmaps. *Bioinformatics* (2022), doi: [10.1093/bioinformatics/btac018](https://doi.org/10.1093/bioinformatics/btac018)
- [M. R. Vollger](#), G. A. Logsdon, P. A. Audano, A. Sulovari, D. Porubsky, P. Peluso, A. M. Wenger, G. T. Concepcion, Z. N. Kronenberg, K. M. Munson, C. Baker, A. D. Sanders, D. C. Spierings, P. M. Lansdorp, U. Surti, M. W. Hunkapiller, E. E. Eichler, Improved assembly and variant detection of a haploid human genome using single-molecule, high-fidelity long reads. *Annals of Human Genetics*. **84**, 125–140 (2019)

- [M. R. Vollger](#) , P. C. Dishuck, M. Sorensen, A. E. Welch, V. Dang, M. L. Dougherty, T. A. Graves-Lindsay, R. K. Wilson, M. J. P. Chaisson, E. E. Eichler, Long-read sequence and assembly of segmental duplications. *Nature Methods*. **16**, 88–94 (2018)
- Corresponding** A. Jha, S. C. Bohaczuk, Y. Mao, J. Ranchalis, B. J. Mallory, A. T. Min, M. O. Hamm, E. Swanson, D. Dubocanin, C. Finkbeiner, T. Li, D. Whittington, W. S. Noble, A. B. Stergachis, [M. R. Vollger](#) , DNA-m6A calling and integrated long-read epigenetic and genetic analysis with fibertools. *Genome Research*, gr.279095.124 (2024)
- Collaborative** K. L. Bubb, M. O. Hamm, J. K. Min, B. Ramirez-Corona, N. A. Mueth, J. Ranchalis, [M. R. Vollger](#) , C. Trapnell, J. T. Cuperus, C. Queitsch, A. B. Stergachis, The regulatory potential of transposable elements in maize (2024), doi: [10.1101/2024.07.10.602892](https://doi.org/10.1101/2024.07.10.602892)
- S. C. Bohaczuk, Z. J. Amador, C. Li, B. J. Mallory, E. G. Swanson, J. Ranchalis, [M. R. Vollger](#) , K. M. Munson, T. Walsh, M. O. Hamm, Y. Mao, A. Lieber, A. B. Stergachis, Resolving the chromatin impact of mosaic variants with targeted Fiber-seq (2024), doi: [10.1101/2024.07.09.602608](https://doi.org/10.1101/2024.07.09.602608)
- W.-W. Liao, M. Asri, J. Ebler, D. Doerr, M. Haukness, G. Hickey, S. Lu, J. K. Lucas, J. Monlong, H. J. Abel, S. Buonaiuto, X. H. Chang, H. Cheng, J. Chu, V. Colonna, J. M. Eizenga, X. Feng, C. Fischer, R. S. Fulton, ... B. Paten, A draft human pangenome reference. *Nature*. **617**, 312–324 (2023)
- W. S. DeWitt, L. Zhu, [M. R. Vollger](#) , M. E. Goldberg, A. Talenti, A. C. Beichman, K. Harris, mutyper: assigning and summarizing mutation types for analyzing germline mutation spectra. *Journal of Open Source Software*. **8**, 5227–5228 (2023)
- D. Porubsky, [M. R. Vollger](#) , W. T. Harvey, A. N. Rozanski, P. Ebert, G. Hickey, P. Hasenfeld, A. D. Sanders, C. Stober, J. O. Korbel, B. Paten, T. Marschall, E. E. Eichler, Gaps and complex structurally variant loci in phased genome assemblies. *Genome Research*. **33**, 496–510 (2023)
- X. Yang, X. Wang, Y. Zou, S. Zhang, M. Xia, [M. R. Vollger](#) , N.-C. Chen, D. J. Taylor, W. T. Harvey, G. A. Logsdon, D. Meng, J. Shi, R. C. McCoy, M. C. Schatz, W. Li, E. E. Eichler, Q. Lu, Y. Mao, A refined characterization of large-scale genomic differences in the first complete human genome (2022), doi: [10.1101/2022.12.17.520860](https://doi.org/10.1101/2022.12.17.520860)
- S. Aganezov, S. M. Yan, D. C. Soto, M. Kirsche, S. Zarate, P. Avdeyev, D. J. Taylor, K. Shafin, A. Shumate, C. Xiao, J. Wagner, J. McDaniel, N. D. Olson, M. E. G. Sauria, [M. R. Vollger](#) , A. Rhie, M. Meredith, S. Martin, J. Lee, ... M. C. Schatz, A complete reference genome improves analysis of human genetic variation. *Science*. **376** (2022), doi: [10.1126/science.abl3533](https://doi.org/10.1126/science.abl3533)
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- A. Gershman, M. E. G. Sauria, X. Guitart, [M. R. Vollger](#) , P. W. Hook, S. J. Hoyt, M. Jain, A. Shumate, R. Razaghi, S. Koren, N. Altomose, G. V. Caldas, G. A. Logsdon, A. Rhie, E. E. Eichler, M. C. Schatz, R. J. O'Neill, A. M. Phillippy, K. H. Miga, W. Timp, Epigenetic patterns in a complete human genome. *Science*. **376** (2022), doi: [10.1126/science.abj5089](https://doi.org/10.1126/science.abj5089)
- S. J. Hoyt, J. M. Storer, G. A. Hartley, P. G. S. Grady, A. Gershman, L. G. de Lima, C. Limouse, R. Halabian, L. Wojenski, M. Rodriguez, N. Altomose, A. Rhie, L. J. Core, J. L. Gerton, W. Makalowski, D. Olson, J. Rosen, A. F. A. Smit, A. F. Straight, ... R. J. O'Neill, From telomere to telomere: The transcriptional and epigenetic state of human repeat elements. *Science*. **376** (2022), doi: [10.1126/science.abk3112](https://doi.org/10.1126/science.abk3112)
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- P. Hsieh, V. Dang, [M. R. Vollger](#) , Y. Mao, T.-H. Huang, P. C. Dishuck, C. Baker, S. Cantsilieris, A. P. Lewis, K. M. Munson, M. Sorensen, A. E. Welch, J. G. Underwood, E. E. Eichler, Evidence for opposing selective forces operating on human-specific duplicated TCAF genes in Neanderthals and humans. *Nature Communications*. **12** (2021), doi: [10.1038/s41467-021-25435-4](https://doi.org/10.1038/s41467-021-25435-4)
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- K. Shafin, T. Pesout, R. Lorig-Roach, M. Haukness, H. E. Olsen, C. Bosworth, J. Armstrong, K. Tigyi, N. Maurer, S. Koren, F. J. Sedlazeck, T. Marschall, S. Mayes, V. Costa, J. M. Zook, K. J. Liu, D. Kilburn, M. Sorensen, K. M. Munson, ... B. Paten, Nanopore sequencing and the Shasta toolkit enable efficient de novo assembly of eleven human genomes. *Nature Biotechnology*. **38**, 1044–1053 (2020)
- A. Sulovari, R. Li, P. A. Audano, D. Porubsky, [M. R. Vollger](#), G. A. Logsdon, W. C. Warren, A. A. Pollen, M. J. P. Chaisson, E. E. Eichler, Human-specific tandem repeat expansion and differential gene expression during primate evolution. *Proceedings of the National Academy of Sciences*. **116**, 23243–23253 (2019)
- P. Hsieh, [M. R. Vollger](#), V. Dang, D. Porubsky, C. Baker, S. Cantsilieris, K. Hoekzema, A. P. Lewis, K. M. Munson, M. Sorensen, Z. N. Kronenberg, S. Murali, B. J. Nelson, G. Chiatante, F. A. M. Maggolini, H. Blanché, J. G. Underwood, F. Antonacci, J.-F. Deleuze, E. E. Eichler, Adaptive archaic introgression of copy number variants and the discovery of previously unknown human genes. *Science*. **366** (2019), doi: [10.1126/science.aax2083](https://doi.org/10.1126/science.aax2083)
- F. A. M. Maggolini, S. Cantsilieris, P. D'Addabbo, M. Manganelli, B. P. Coe, B. L. Dumont, A. D. Sanders, A. W. C. Pang, [M. R. Vollger](#), O. Palumbo, P. Palumbo, M. Accadia, M. Carella, E. E. Eichler, F. Antonacci, Genomic inversions and GOLGA core duplicons underlie disease instability at the 15q25 locus. *PLOS Genetics*. **15**, e1008075 (2019)

Presentations

Fiber-seq and tools to understand the regulatory genome in a disease context

European Society of Human Genetics (ESHG)

May 2025

Allianz MiCo in Milan, Italy

Fiber-seq Inferred Regulatory Elements with diploid T2T genomes

Telomere-to-telomere face-to-face conference

Summer 2024

University of California Santa Cruz

Comprehensive diploid genetic and epigenetic profiles with single-molecule precision

Division of Medical Genetics Seminar Series

April 2023

University of Washington

Comprehensive diploid genetic and epigenetic profiles with single-molecule precision

AGBT 2023

Feb 2023

Hollywood, Florida

A complete view of segmental duplications and their variation

Genome Sciences 20th anniversary symposium

Dec 2022

University of Washington

Using a complete human reference to explore variation in segmental duplications

Long-Read, Long-Range scientific interest group

Oct 2022

NHGRI, remote

Increased mutation rate and interlocus gene conversion within human segmental duplications

Telomere-to-telomere face-to-face conference

Aug 2022

University of California Santa Cruz

Segmental duplications and their variation in a complete human genome

UCSC BME departmental seminar series

March 2022

University of California Santa Cruz, remote

Segmental duplications and their variation in a complete human genome

NHGRI computational biology seminar series

Oct 2021

NHGRI, remote

A complete view of segmental duplications and their variation

American Society of Human Genetics, Section talk

Sep 2021

remote

A complete view of segmental duplications and their variation

T2T and HPRC conference

Sep 2020

University of Washington

Improved Assembly of Segmental Duplications Using HiFi

Pacific Biosciences User Group Meeting

Sep 2019

University of Delaware

Teaching Experience

Gene discovery and comparative genomics

Invited Lecture, Genomics and Proteomics, undergraduate course

October 2022

University of Washington

Introduction to Statistical Genomics

Primary Instructor, Introduction to Statistical Genomics, graduate course

Spring 2022

University of Washington

Introduction to Computational Molecular Biology

Teaching Assistant, Lead weekly discussion sections, organized and graded assignments, and held office hours

Winter 2020

University of Washington

Fundamentals of Genetics and Genomics

Teaching Assistant, Lead weekly discussion sections, organized and graded assignments, and held office hours

Summer 2019

University of Washington

Programming Languages

Daily Use Rust | Python | R | Snakemake | Bash
As needed C++ | LaTeX | typst

Professional Organizations

2023-Present Somatic Mosaicism Across Human Tissues consortium (SMaHT)
2021-Present American Society of Human Genetics (ASHG)
2020-Present Telomere to Telomere consortium (T2T)
2020-Present Human Pangenome Reference Consortium (HPRC)

References

Advisor [Andrew B. Stergachis](#) | absterga@uw.edu
Advisor [Evan E. Eichler](#) | eee@gs.washington.edu
Collaborator [Adam Phillippy](#) | adam.phillippy@nih.gov
Collaborator [William Noble](#) | wnoble@uw.edu
Collaborator [Winston Timp](#) | wtimp@jhu.edu