# 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

Dissertation: Assembly of segmental duplications and their variation in humans

· Advisor: Evan E. Eichler

• Completed the Advanced Data Sience Option

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

Departments of Computer Science and Quantitative and Computational Biology

· Student of the Integrated Science Curriculum

Certificate in Quantitative and Computational Biology

#### Associate of Arts Degrees at College of the Redwoods

AA in Mathematics | AA in Science

Sep. 2008 - June 2011

Sep. 2011 - June 2015

Princeton, New Jersey

Sep 2016 - March 2021

Seattle, Washington

Eureka, California

#### **Postdoctoral Experience**

Postdoctoral Scholar in the Division of Medical Genetics

In the lab of Andrew B. Stergachis

April 2022 - Present

University of Washington

Postdoctoral Scholar in the Department of Genome Sciences

In the lab of Evan E. Eichler

March 2021 - April 2022 University of Washington

## Funding and Awards\_

K99/R00 Pathway to Independence Award

National Institute of General Medical Sciences, 1K99GM155552-01

NIH/NHGRI T32 Genome Training Grant

Division of Medical Genetics at University of Washington

BDGN, Big Data in Genomics and Neuroscience

Genome Sciences at University of Washington

NIH/NHGRI T32 Genome Training Grant

Genome Sciences at University of Washington

Summer 2024 - present

Seattle, Washington

Fall 2022 - Fall 2024

Seattle, Washington

Fall 2017 - Fall 2019

Seattle, Washington

Fall 2016 - Fall 2017

Seattle, Washington

#### **Publications**\_

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

- 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
- 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
- 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
- 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)

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- 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
  - 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 Fiberseq (2024), doi: 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, <u>M. R. Vollger</u>, 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
  - 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
  - N. Altemose, G. A. Logsdon, A. V. Bzikadze, P. Sidhwani, S. A. Langley, G. V. Caldas, S. J. Hoyt, L. Uralsky, F. D. Ryabov, C. J. Shew, M. E. G. Sauria, M. Borchers, A. Gershman, A. Mikheenko, V. A. Shepelev, T. Dvorkina, O. Kunyavskaya, M. R. Vollger, A. Rhie, ... K. H. Miga, Complete genomic and epigenetic maps of human centromeres. *Science*. 376 (2022), doi: 10.1126/science.abl4178
  - 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. Altemose, 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
  - 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. Altemose, 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
  - S. Nurk, S. Koren, A. Rhie, M. Rautiainen, A. V. Bzikadze, A. Mikheenko, M. R. Vollger, N. Altemose, L. Uralsky, A. Gershman, S. Aganezov, S. J. Hoyt, M. Diekhans, G. A. Logsdon, M. Alonge, S. E. Antonarakis, M. Borchers, G. G. Bouffard, S. Y. Brooks, ... A. M. Phillippy, The complete sequence of a human genome. *Science*. 376, 44–53 (2022)
  - 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
  - G. A. Logsdon, M. R. Vollger, P. Hsieh, Y. Mao, M. A. Liskovykh, S. Koren, S. Nurk, L. Mercuri, P. C. Dishuck, A. Rhie, L. G. de Lima, T. Dvorkina, D. Porubsky, W. T. Harvey, A. Mikheenko, A. V. Bzikadze, M. Kremitzki, T. A. Graves-Lindsay, C. Jain, ... E. E. Eichler, The structure, function and evolution of a complete human chromosome 8. *Nature*. 593, 101–107 (2021)
  - D. Porubsky, , P. Ebert, P. A. Audano, M. R. Vollger, W. T. Harvey, P. Marijon, J. Ebler, K. M. Munson, M. Sorensen, A. Sulovari, M. Haukness, M. Ghareghani, P. M. Lansdorp, B. Paten, S. E. Devine, A. D. Sanders, C. Lee, M. J. P. Chaisson, ... T. Marschall, Fully phased human genome assembly without parental data using single-cell strand sequencing and long reads. *Nature Biotechnology* (2020), doi: 10.1038/s41587-020-0719-5
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  - G. A. Logsdon, M. R. Vollger, E. E. Eichler, Long-read human genome sequencing and its applications. *Nature Reviews Genetics*. 21, 597–614 (2020)

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- S. Nurk, B. P. Walenz, A. Rhie, M. R. Vollger, G. A. Logsdon, R. Grothe, K. H. Miga, E. E. Eichler, A. M. Phillippy, S. Koren, HiCanu: accurate assembly of segmental duplications, satellites, and allelic variants from high-fidelity long reads. Genome Research. 30, 1291-1305 (2020)
- K. H. Miga, S. Koren, A. Rhie, M. R. Vollger, A. Gershman, A. Bzikadze, S. Brooks, E. Howe, D. Porubsky, G. A. Logsdon, V. A. Schneider, T. Potapova, J. Wood, W. Chow, J. Armstrong, J. Fredrickson, E. Pak, K. Tigyi, M. Kremitzki, ... A. M. Phillippy, Telomere-to-telomere assembly of a complete human X chromosome. *Nature*. **585**, 79–84 (2020)
- 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. Maggiolini, 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
- F. A. M. Maggiolini, 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)

Fiber-seq Inferred Regulatory Elements with diploid T2T genomes

Telomere-to-telomere face-to-face conference

Comprehensive diploid genetic and epigenetic profiles with single-molecule precision

Division of Medical Genetics Seminar Series

Comprehensive diploid genetic and epigenetic profiles with single-molecule precision

A complete view of segmental duplications and their variation

Segmental duplications and their variation in a complete human genome

Segmental duplications and their variation in a complete human genome

Genome Sciences 20th anniversary symposium

Using a complete human reference to explore variation in segmental duplications

Long-Read, Long-Range scientific interest group

Increased mutation rate and interlocus gene conversion within human segmental duplications Telomere-to-telomere face-to-face conference

UCSC BME departmental seminar series

NHGRI computational biology seminar series

A complete view of segmental duplications and their variation American Society of Human Genetics, Section talk

A complete view of segmental duplications and their variation T2T and HPRC conference

Improved Assembly of Segmental Duplications Using HiFi

Pacific Biosciences User Group Meeting

Allianz MiCo in Milan, Italy Summer 2024

University of California Santa Cruz

April 2023

University of Washington

Feb 2023

Hollywood, Florida

May 2025

Dec 2022

University of Washington

Oct 2022 NHGRI, remote

Aug 2022

University of California Santa Cruz

March 2022

University of California Santa Cruz, remote

Oct 2021

NHGRI, remote

Sep 2021

Sep 2020

University of Washington

Sep 2019

University of Delaware

# **Teaching Experience**

Gene discovery and comparative genomics

Invited Lecutre, Genomics and Proteomics, undergraduate course

**Introduction to Statistical Genomics** 

Primary Instructor, Introduction to Statistical Genomics, graduate course

Introduction to Computational Molecular Biology

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

**Fundamentals of Genetics and Genomics** 

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

October 2022

University of Washington

Spring 2022

University of Washington

Winter 2020

University of Washington

Summer 2019

University of Washington

Mitchell R Vollger Curriculum vitae

# **Programming Languages**\_

```
Daily Use Rust | Python | R | Snakemake | Bash
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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 | <u>absterga@uw.edu</u>
Advisor Evan E. Eichler | <u>eee@gs.washington.edu</u>
Collaborator Adam Phillippy | <u>adam.phillippy@nih.gov</u>

Collaborator William Noble | wnoble@uw.edu Collaborator Winston Timp | wtimp@jhu.edu

Mitchell R. Vollger Curriculum vitae