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

Seattle, Washington

Sep 2016 - March 2021

- · Advisor: Evan E. Eichler
- Completed the Advanced Data Science Option

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

Departments of Computer Science and Quantitative and Computational Biology

Sep. 2011 - June 2015 Princeton, New Jersey

- 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

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

NIH/NHGRI T32 Genome Training Grant

BDGN, Big Data in Genomics and Neuroscience

Genome Sciences at University of Washington

Genome Sciences at University of Washington

Summer 2024 - present University of Washington

Fall 2022 - Fall 2024

University of Washington

Fall 2017 - Fall 2019

University of Washington

Fall 2016 - Fall 2017

University of Washington

Publications

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, Genome Research, in press, doi: 10.1101/

gr.279095.124.

First Author M. R. Vollger, J. Korlach, K. C. Eldred, E. Swanson, J. G. Underwood, S. C. Bohaczuk, Y. Mao, Y.-H. H. Cheng, J.

Ranchalis, 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, ... A. B. Stergachis, Nature Genetics, in press, doi: 10.1038/s41588-024-02067-0.

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, 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, *Nature*, in press, doi: <u>10.1038/s41586-023-05895-y</u>.

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.

Mitchell R. Vollger Curriculum vitae

- 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, *Annals of Human Genetics*, in press, doi: 10.1111/ahg.12364
- 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, *Nature Methods*, in press, doi: 10.1038/s41592-018-0236-3.
- Collaborative T. D. Real, P. Hebbar, D. Yoo, F. Antonacci, I. Pačar, M. Diekhans, G. J. Mikol, O. G. Popoola, B. J. Mallory, M. R. Vollger, P. C. Dishuck, X. Guitart, A. N. Rozanski, K. M. Munson, K. Hoekzema, J. E. Ranchalis, S. J. Neph, A. E. Sedeño-Cortes, B. Paten, ... E. E. Eichler, Genetic diversity and regulatory features of human-specificNOTCH2NLduplications (2025), doi: 10.1101/2025.03.14.643395
 - D. Dubocanin, A. Kalygina, J. M. Franklin, C. Chittenden, <u>M. R. Vollger</u>, S. Neph, A. B. Stergachis, N. Altemose,
 Integrating Single-Molecule Sequencing and Deep Learning to Predict Haplotype-Specific 3D Chromatin Organization in a Mendelian Condition (2025), doi: <u>10.1101/2025.02.26.640261</u>.
 - E. G. Swanson, Y. Mao, B. J. Mallory, M. R. Vollger, J. Ranchalis, S. C. Bohaczuk, N. L. Parmalee, J. T. Bennett, A. B. Stergachis, Deaminase-assisted single-molecule and single-cell chromatin fiber sequencing (2024), doi: 10.1101/2024.11.06.622310.
 - 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, <u>M. R. Vollger</u>, 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, *Nature*, in press, doi: 10.1038/s41586-023-05896-x.
 - W. S. DeWitt, L. Zhu, M. R. Vollger, M. E. Goldberg, A. Talenti, A. C. Beichman, K. Harris, *Journal of Open Source Software*, in press, doi: 10.21105/joss.05227.
 - 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, *Genome Research*, in press, doi: 10.1101/gr.277334.122.
 - 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, *Science*, in press, doi: 10.1126/science.abj6987.
 - 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, *Nature*, in press, doi: 10.1038/s41586-021-03420-7.
 - D. Porubsky, , P. Ebert, P. A. Audano, <u>M. R. Vollger</u>, 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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- W. C. Warren, R. A. Harris, M. Haukness, I. T. Fiddes, S. C. Murali, J. Fernandes, P. C. Dishuck, J. M. Storer, M. Raveendran, L. W. Hillier, D. Porubsky, Y. Mao, D. Gordon, M. R. Vollger, A. P. Lewis, K. M. Munson, E. DeVogelaere, J. Armstrong, M. Diekhans, ... E. E. Eichler, *Science*, in press, doi: 10.1126/science.abc6617.
- G. A. Logsdon, M. R. Vollger, E. E. Eichler, Nature Reviews Genetics, in press, doi: 10.1038/s41576-020-0236-x.
- 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, *Genome Research*, in press, doi: 10.1101/gr.263566.120.
- 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, *Nature*, in press, doi: 10.1038/s41586-020-2547-7.
- 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, *Nature Biotechnology*, in press, doi: 10.1038/s41587-020-0503-6.
- 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, *Proceedings of the National Academy of Sciences*, in press, doi: 10.1073/pnas.1912175116.
- 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, *PLOS Genetics*, in press, doi: 10.1371/journal.pgen.1008075.

Presentations

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

May 2025
Allianz MiCo in Milan, Italy

European Society of Human Genetics (ESHG)

Oct 2024

Computational tools for Fiber-seq and Fiber-seq Inferred Regulatory Elements

Seattle Children's Research Institute

BBI Long-read Symposium

Aug 2024

Fiber-seq Inferred Regulatory Elements with diploid T2T genomes

University of California Santa Cruz

Telomere-to-telomere face-to-face conference

Apr 2023

Comprehensive diploid genetic and epigenetic profiles with single-molecule precision Division of Medical Genetics Seminar Series

University of Washington

Comprehensive diploid genetic and epigenetic profiles with single-molecule precision

Feb 2023

AGBT 2023

Hollywood, Florida

A complete view of segmental duplications and their variation

Dec 2022
University of Washington

Genome Sciences 20th anniversary symposium

Using a complete human reference to explore variation in segmental duplications Long-Read, Long-Range scientific interest group

NHGRI, remote

Oct 2022

Increased mutation rate and interlocus gene conversion within human segmental duplications

Aug 2022

Telomere-to-telomere face-to-face conference

University of California Santa Cruz

Segmental duplications and their variation in a complete human genome

Mar 2022

UCSC BME departmental seminar series

University of California Santa Cruz, remote

Segmental duplications and their variation in a complete human genome

Oct 2021

NHGRI computational biology seminar series

NHGRI, remote

A complete view of segmental duplications and their variation

Sep 2021 remote

American Society of Human Genetics, Section talk

Sep 2020

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

University of Washington

Improved Assembly of Segmental Duplications Using HiFi

Sep 2019

Pacific Biosciences User Group Meeting

 $University\ of\ Delaware$

Teaching Experience

Gene discovery and comparative genomics

October 2022

Invited Lecture, Genomics and Proteomics, undergraduate course

University of Washington

Spring 2022

Introduction to Statistical Genomics

University of Washington

Primary Instructor, Introduction to Statistical Genomics, graduate course

Introduction to Computational Molecular Biology

Winter 2020 University of Washington

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

Summer 2019

Fundamentals of Genetics and Genomics

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_

AdvisorAndrew B. Stergachis| absterga@uw.eduAdvisorEvan E. Eichler| eee@gs.washington.eduCollaboratorAdam Phillippy| adam.phillippy@nih.govCollaboratorWilliam Noble| wnoble@uw.eduCollaboratorWinston Timp| wtimp@jhu.edu

Mitchell R. Vollger Curriculum vitae