Mitchell R. **Vollger**

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0000-0002-8651-1615 | Ph.D.

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 Sience 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

— **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)

—	**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 Fiber-seq (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, **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
—	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
—	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, Sequence diversity analyses of an improved rhesus macaque genome enhance its biomedical utility. *Science*. **370**, eabc6617 (2020)
—	G. A. Logsdon, **M. R. Vollger** , E. E. Eichler, Long-read human genome sequencing and its applications. *Nature Reviews Genetics*. **21**, 597–614 (2020)

— 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 — *May 2025*
European Society of Human Genetics (ESHG) — *Allianz MiCo in Milan, Italy*

Fiber-seq Inferred Regulatory Elements with diploid T2T genomes — *Summer 2024*
Telomere-to-telomere face-to-face conference — *University of California Santa Cruz*

Comprehensive diploid genetic and epigenetic profiles with single-molecule precision — *April 2023*
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*
Genome Sciences 20th anniversary symposium — *University of Washington*

Using a complete human reference to explore variation in segmental duplications — *Oct 2022*
Long-Read, Long-Range scientific interest group — *NHGRI, remote*

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 — *March 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*
American Society of Human Genetics, Section talk — *remote*

A complete view of segmental duplications and their variation — *Sep 2020*
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 Lecutre, Genomics and Proteomics, undergraduate course — *University of Washington*

Introduction to Statistical Genomics — *Spring 2022*
Primary Instructor, Introduction to Statistical Genomics, graduate course — *University of Washington*

Introduction to Computational Molecular Biology — *Winter 2020*
Teaching Assistant, Lead weekly discussion sections, organized and graded assignments, and held weekly office hours — *University of Washington*

Fundamentals of Genetics and Genomics — *Summer 2019*
Teaching Assistant, Lead weekly discussion sections, organized and graded assignments, and held weekly office hours — *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
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