Harriet Dashnow, Ph.D.

Assistant Professor, University of Colorado Empowering genetic diagnostics through bioinformatics

Education

Institution	Degree	Completed	Field of study
The University of Melbourne	PhD	Nov 2019	Bioinformatics
The University of Melbourne	MSc	Dec 2013	Bioinformatics (Dean's honours)
The University of Melbourne	BSc	Dec 2011	Genetics, Biochemistry and Molecular Biology
The University of Melbourne	BA	Dec 2011	Psychology

Summary

I develop computational methods and workflows to increase the diagnosis of rare diseases with a special focus on Tandem Repeat (TR) loci. I partner with clinicians and researchers to analyze patient genomes at scale and establish genetic diagnoses.

Research Direction

- Establish a genetic diagnosis for individuals with rare diseases, directly and by enabling others.
- Develop computational methods for genetic variant discovery and interpretation.
- Emphasis on tandem repeats and other complex genetic variants.
- Understand population variation to accelerate the discovery of new TR disease loci.
- Develop computational methods to fully utilize new technolgies such as long-read sequencing.

Funding

Current:

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NIH NGHRI K99/R00 Pathway to Independence Award – USD $1,184,940/5 years, Feb 2023–Jan 2028
NHMRC Investigator Award (EL1) – AUD $662,040/5 years, July 2024–Jun 2029 (St Vincent's Institute)
Past:
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NIH NGHRI T32 in Genomic Medicine – USD \$76,196/year, 2022–2023 University of Utah Postdoc Travel Assistance Award – USD \$500, 2022 NIH NHLBI BioData Catalyst Fellowship – USD \$69,733, 2020 Australian Genomics Health Alliance PhD top-up scholarship – AUD \$10,000/year, 2017–2018 MCRI PhD Top Up Scholarship – AUD \$5,000/year, 2015–2018 Australian Postgraduate Award/Research Training Program – ~AUD \$26,000/year, 2015–2018 Australasian Genomic Technologies Association Small Grants scheme – AUD \$5,000, 2014 Victorian Life Science Computational Initiative Travel Award AUD – \$3,000, 2014 Victorian Life Science Computational Initiative Bursary – AUD \$5,000, 2012 Cancer Therapeutics (CTx) Summer Scholarship – AUD \$1,500, 2011 MCRI Summer Student Scholarship – AUD \$1,500, 2008

Research Experience

Jul 2024 –	Assistant Professor			
present	Department of Biomedical Informatics, University of Colorado Anschutz Medical Campus, USA			
Jun 2019 – Jun 2024	Postdoctoral Research Associate, Aaron Quinlan Lab			
	Eccles Institute of Human Genetics, The University of Utah, USA			
Mar 2015 –	PhD Student/Bioinformatician, Alicia Oshlack Lab			
Jun 2019	Murdoch Children's Research Institute, The Royal Children's Hospital, Melbourne, Australia			
Nov – Dec 2015	Visiting Scholar, Daniel McArthur Lab			
	Broad Institute of MIT and Harvard/Massachusetts General Hospital, Massachusetts, USA			
Dec 2013 – Feb 2015	Bioinformatician, Life Science Computation Centre, lead by Andrew Lonie			
	Melbourne Genomic Health Alliance and Victorian Life Sciences Computation Initiative (now Melbourne Bioinformatics), The University of Melbourne, Australia			
Dec 2012 –	Research Assistant, Kathryn Holt's Lab			
Jul 2013	Department of Biochemistry and Molecular Biology, The University of Melbourne, Australia			
	Initially funded by the Victorian Life Sciences Computation Initiative Internship			

Nov 2010 – Nov 2011	Undergraduate research placement (UROP), Brodnicki Lab St Vincent's Institute, St Vincent's Hospital, Melbourne, Australia Genetics of type I diabetes in mouse models
Jan 2008 – Dec 2010	Technical Assistant, Cell and Gene Therapy (Heidi Peters)
	Murdoch Children's Research Institute, The Royal Children's Hospital, Melbourne, Australia
	Developed and characterised humanized genetic mouse models for methylmalonic aciduria.

Teaching Experience

- Nov 2021 **Teaching Assistant**, Advanced Sequencing Technologies & Applications Cold Spring Harbor Laboratory, New York, USA
- 2014–2022 Instructor and lesson developer. Python, R, Unix, Bash, Git.

The Carpentries (Software Carpentry and Data Carpentry).

Developed and maintained several internationally used curricula on R and data analysis. I have taught the following workshops. For many, I was invited (with travel funded by the host):

- Oct 2022 University of Utah Unix, Python, Git
- Jul 2021 University of Utah Unix, Python, R
- Apr 2018 Walter and Eliza Hall Institute, Melbourne Unix, R, Git
- Apr 2017 Walter and Eliza Hall Institute, Melbourne Unix, R, Git
- June 2016 University of Melbourne Unix, R, Git
- Nov 2015 Women in Science & Engineering, New York University Unix, Python, Git, R
- Oct 2015 University of Sydney SQL, R, spreadsheets
- May 2015 Murdoch Childrens Research Institute Unix, Git, R
- Apr 2015 Biosciences, University of Melbourne Unix, Git, R, Python
- Mar 2015 Indian Statistical Institute, Bangalore SQL, R, spreadsheets
- Dec 2014 University of Melbourne R, Unix, Git

2017 **Book co-author.** Elegant SciPy: The art of scientific python.

O'Reilly Media, Inc.

The book teaches scientific Python to researchers. It has sold more than 9,500 copies and has been translated into three languages.

2017 Instructor and lesson developer. RNAseq analysis in R

COMBINE/Galaxy Training Network

Developed curriculum materials on R and RNAseq analysis. These formed the basis for the current Galaxy Training Network RNAseq curriculum.

2013–2017 Lead demonstrator, Genomics and Bioinformatics (coordinator: Leanne Tilly)
 Department of Biochemistry and Molecular Biology, The University of Melbourne, Australia
 Developed and marked assignments, and led computational labs. Graduate-level course.

2014–2015	Teaching Assistant, Algorithms for Functional Genomics (coordinator: Clare Sloggett)
	Computer Science, The University of Melbourne, Australia
	Graduate-level course teaching fundamental algorithms in bioinformatics.
Mar 2014	Guest lecturer, Kwong Lee Dow Young Scholars Case Competition
	The University of Melbourne, Australia
	Developed materials, lectured genomics and personalised medicine to hundreds of high-school students. Facilitated discussion of the ethical and financial cases for clinical sequencing.
Feb 2012 – Dec 2013	College tutor in residence, Genetics, Biochemistry and Molecular Biology, Biomedicine
	St Hilda's College, The University of Melbourne, Australia
	Small class and individual tutoring of undergraduate students. Pastoral support and mentoring.
2011–2013	College tutor, Genetics, Biochemistry and Molecular Biology, Biomedicine
	Trinity College, The University of Melbourne, Australia
	Small class and individual tutoring of undergraduate students.
2007–2013	Private Tutor
	Self Employed, Melbourne, Australia
	Tutoried undergraduate Biology, high-school English, Biology, Mathematics and Chemistry

Awards and Prizes

Australian Bioinformatics and Computational Biology Society 2020 - **Outstanding PhD thesis award** American Society of Human Genetics 2020 - Reviewers' Choice poster abstract award COMBINE Symposium, Adelaide 2017 – **winner best talk** GeneMappers, Geelong 2017 – **winner best student talk** Lorne Genome 2017 – **winner student poster prize, invited speaker** in the Bioinformatics workshop International Society for Computational Biology, Boston, USA 2014 - **winner best student talk**

Selected Publications

For a full list of publications, please see my profile on Google Scholar

- 1. Hiatt, L., Weisburd, B., Dolzhenko, E., VanNoy, G.E., Kurtas, E.N., Rehm, H.L., Quinlan, A. and **Dashnow, H.**, 2024. STRchive: a dynamic resource detailing population-level and locus-specific insights at tandem repeat disease loci. *medRxiv* doi: https://doi.org/10.1101/2024.05.21.24307682.
- 2. Gustafson, J.A., ... Dashnow, H., ..., Miller, D.E. (2024). Nanopore sequencing of 1000 Genomes

Project samples to build a comprehensive catalog of human genetic variation. *medRxiv* doi: https://doi.org/10.1101/2024.03.05.24303792.

- 3. Tanudisastro, H. A., Deveson, I. W., **Dashnow, H.**[∞], & MacArthur, D. G.[∞] (2024). Advances in sequencing and characterising short tandem repeats (STRs) in the human genome. *Nature Reviews Genetics*, 1-16. ∞ Corresponding authors.
- Dolzhenko, E.[#], English, A.[#], Dashnow, H.[#], De Sena Brandine, G., Mokveld, T., Rowell, W. J., ... & Eberle, M. A. (2024). Characterization and visualization of tandem repeats at genome scale. *Nature Biotechnology*. # These authors contributed equally.
- Dashnow, H., Pedersen, B. S., Hiatt, L., Brown, J., Beecroft, S. J., Ravenscroft, G., ... & Quinlan, A. R. (2023). STRling: a k-mer counting approach that detects short tandem repeat expansions at known and novel loci. *Genome Biology*, 23(1), 1-20.
- Pedersen, B. S., Brown, J. M., Dashnow, H., Wallace, A. D., Velinder, M., Tristani-Firouzi, M., ... & Quinlan, A. R. (2021). Effective variant filtering and expected candidate variant yield in studies of rare human disease. *NPJ Genomic Medicine*, 6(1), 1-8.
- 7. Shere, H., Weijer, L., **Dashnow, H.**, Moreno, L. E., Foxworthy Scott, S., & Baker, H. (2021). Chronic Lactation Insufficiency Is a Public Health Issue. *Breastfeeding Medicine*, 16(12), 933-934.
- Georgeson, P., Syme, A., Sloggett, C., Chung, J., Dashnow, H., Milton, M., ... & Pope, B. (2019). Bionitio: demonstrating and facilitating best practices for bioinformatics command-line software. *GigaScience*, 8(9), giz109.
- 9. **Dashnow, H.**, Bell, K. M., Stark, Z., Tan, T. Y., White, S. M., & Oshlack, A. (2019). Pooled-parent exome sequencing to prioritise de novo variants in genetic disease. *bioRxiv*, 601740.
- Dashnow, H., Lek, M., Phipson, B., Halman, A., Sadedin, S., Lonsdale, A., ... & Oshlack, A. (2018). STRetch: detecting and discovering pathogenic short tandem repeat expansions. *Genome biology*, 19(1), 1-13.
- 11. Nunez-Iglesias, J., Van Der Walt, S., & **Dashnow, H.** (2017). *Elegant SciPy: The art of scientific python*. " O'Reilly Media, Inc.".
- 12. Stark, Z., **Dashnow, H.**, Lunke, S., Tan, T. Y., Yeung, A., Sadedin, S., ... & James, P. A. (2017). A clinically driven variant prioritization framework outperforms purely computational approaches for the diagnostic analysis of singleton WES data. *European Journal of Human Genetics*, *25*(11), 1268-1272.
- 13. Lonsdale, A., Sietsma Penington, J., Rice, T., Walker, M., & **Dashnow, H.** (2016). Ten simple rules for a bioinformatics journal club. *PLOS Computational Biology*, *12*(1), e1004526.
- Sadedin, S. P., Dashnow, H., James, P. A., Bahlo, M., Bauer, D. C., Lonie, A., ... & Thorne, N. P. (2015). Cpipe: a shared variant detection pipeline designed for diagnostic settings. *Genome medicine*, 7(1), 1-10.
- 15. **Dashnow, H.**, Lonsdale, A., & Bourne, P. E. (2014). Ten simple rules for writing a PLOS ten simple rules article. *PLOS Computational Biology*, *10*(10), e1003858.
- 16. Kowsar, Y., **Dashnow, H.**, & Lonie, A. (2014, December). Data Interlocking: Coupling analytics to the data. In *2014 IEEE/ACM 7th International Conference on Utility and Cloud Computing* (pp.

696-701). IEEE.

- Inouye, M., Dashnow, H., Raven, L. A., Schultz, M. B., Pope, B. J., Tomita, T., ... & Holt, K. E. (2014). SRST2: rapid genomic surveillance for public health and hospital microbiology labs. *Genome medicine*, 6(11), 1-16.
- Buck, N. E., Dashnow, H., Pitt, J. J., Wood, L. R., & Peters, H. L. (2012). Development of transgenic mice containing an introduced stop codon on the human methylmalonyl-CoA mutase locus. *PLOS ONE*.

Selected Presentations (*upcoming)

Biomolecular Horizons, Melbourne, Australia, Sept 2024 - invited international speaker*

University of Utah Center for Genomic Medicine and the Primary Children's Hospital Center for Personalized Medicine Symposium, May 2024 - poster

Broad Institute Structural Variation Seminar, March 2024 - invited seminar

American Society of Human Genetics, Nov 2023 - platform talk

DELPHI Data Science Symposium, Oct 2023 - poster

Stanford Genetics Conference on Structural Variants and DNA Repeats, Sept 2023 - talk

XXIIIrd International Congress of Genetics, Melbourne, Australia, July 2023 - invited international speaker

University of Utah Center for Genomic Medicine and the Primary Children's Hospital Center for Personalized Medicine Symposium, May 2023 - invited speaker

NHGRI Training Meeting, Salt Lake City, Utah, April 2023 - poster

ACMG Annual Clinical Genetics Meeting, Salt Lake City, Utah, March 2023 - poster

Northwestern University, Feinberg School of Medicine, Neurology, March 2023 - invited seminar

University of Colorado Anschutz, Department of Biomedical Informatics, Feb 2023 - invited seminar

Murdoch Children's Research Institute, Australia, Dec 2022 - invited seminar

University of Utah, Human Genetics: Department Retreat, Nov 2022 - talk

Stanford Genetics Conference on Structural Variants and DNA Repeats, Sept 2022 - talk

Undiagnosed Diseases Network Tool Building Coalition, June 2022 - invited seminar

Biology of Genomes 2022, NY, USA - poster

TOPMed Structural Variant Working Group, December 2021 - invited seminar

BioData Catalyst Quarterly Meeting, December 2021 - invited seminar

American Society of Human Genetics 2020 - poster, Reviewers' Choice poster abstract award

TOPMed Structural Variant Working Group, July 2020 - invited seminar

University of Utah, Human Genetics: Research in Progress seminar, June 2020

Biology of Genomes 2020, virtual - poster

Undiagnosed Diseases Network Tool Building Coalition, February 2020 - invited seminar

Center for Genomic Medicine Symposium, University of Utah, January 2020 - poster

Genome Sciences, University of Washington, December 2019 - invited seminar

Genome Informatics 2019, NY, USA - selected talk

Sanger Institute, Cambridge, UK, March 2018 - invited seminar

Genomics of Rare Disease, Cambridge, UK 2018 - invited international speaker

Big Data Institute, Oxford, UK, March 2018 - invited seminar

Imperial College, London, UK, March 2018 - invited seminar

Center for Mendelian Genomics, University of Washington, January 2018 - invited seminar

Australian Bioinformatics and Computational Biology Association, Adelaide 2017 - talk

COMBINE Symposium, Adelaide 2017 - winner best talk

GeneMappers, Geelong 2017 – winner best student talk

Lorne Genome 2017 - winner student poster prize, invited speaker in the Bioinformatics workshop

Best Practices in Bioinformatics Training, Brisbane 2016 - invited speaker

Australian Bioinformatics and Computational Biology Association, Brisbane 2016 - talk

ISI-CODATA Bangalore, India 2015 - invited international speaker

International Society for Computational Biology ISMB, Boston, USA 2014 - winner best student talk

InCoB, Sydney 2014 - talk and poster

COMBINE Symposium 2013 - talk

Bio21 Undergraduate Research Opportunities Conference, Melbourne 2011 - talk

Service to the Scientific Community

Established the Human Genetics Postdoc Representative position and Seminar Series, University of Utah, Nov 2023–March 2024

K99 peer mentoring group 2021-present

Utah Postdoctoral Association Board 2020-2024

- Advocacy Committee Chair 2020–2024
- Senior Chair 2021–2022

• Junior Chair 2020–2021

University of Utah Distinguished Mentor Award Selection Committee 2022

Keynote seminar organizer: Claus O Wilke, National Postdoc. Appreciation Week, Sept 2022

American Society of Human Genetics 2021 session chair "Identifying repeats and structural variants underlying complex phenotypes".

University of Utah Academic Senate Postdoc Policy Committee 2020-2021

Australian Bioinformatics and Computational Biology Society Executive Committee - Student Representative 2016–2017

COMBINE (The Australian Bioinformatics and Computational Biology Student Association)

- Student Representative to ABACBS 2016–2017
- President 2015
- Vice-President/Secretary 2014

COMBINE Symposium Sydney 2015 - Conference convener

COMBINE Symposium Melbourne 2014 - Conference organizing committee

Parkville Bioinformatics Journal Club - Co-organizer 2013-2017

International Society for Computational Biology Student Council, Communications Committee 2014

Bio21 Undergraduate Research Opportunities Conference, Melbourne 2012, Bioinformatics session chair

Bio21 Undergraduate Research Opportunities Committee 2011-2014

I review for several journals, including Bioinformatics, PLOS Computational Biology, American Journal of Human Genetics, and Genome Medicine.

Professional Memberships

Genetics Society of America 2023-present

American Society of Human Genetics 2020-present

Australian Bioinformatics and Computational Biology Society 2014-present

National Postdoctoral Association 2020-2024

Utah Postdoctoral Association 2020-2024

COMBINE (The Australian Bioinformatics and Computational Biology Student Association) 2013–2017

International Society for Computational Biology 2014–2016

Australasian Genomic Technologies Association (AGTA) 2014–2015

In addition to my scientific contributions, I also strongly believe in contributing to the broader research community through teaching and community outreach. To this end I have volunteered to teach Software and Data Carpentry workshops across Australia, in the US, and in India, to upskill researchers in computational approaches. I have developed lesson materials for general data analysis as well as genomics and RNAseq. I have substantial experience in teaching bioinformatics and genetics at both the undergraduate and graduate levels. I co-authored the book "Elegant SciPy", which teaches cross-disciplinary approaches to scientific programming. I have also written editorials for PLoS Computational Biology and volunteered on a number of scientific and conference committees (detailed below).

- Nunez-Iglesias, J., van der Walt, S., & Dashnow, H. Elegant SciPy: The Art of Scientific Python. (2017), O'Reilly.
- Dashnow, H., Lonsdale, A., & Bourne, P. E. (2014). Ten simple rules for writing a PLOS ten simple rules article. PLoS Computational Biology, 10(10), e1003858.
- Lonsdale, A., ..., & Dashnow, H. (2016). Ten simple rules for a bioinformatics journal club. PLoS computational biology, 12(1), e1004526.

I believe that **diversity is key to a successful research community**. To that end, I committed to mentoring and supporting women, members of the LGBQT+ community, neurodivergent people, people of colour, and parents. I further commit to making our research spaces, such as conferences, more inclusive of the full spectrum of human diversity.