# Amy L. Williams

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| EDUCATION | **Massachusetts Institute of Technology**, Cambridge, Massachusetts, USA |

Ph.D., Computer Science **February 2010**
- Advisors: Professors David K. Gifford and David E. Housman

S.M., Electrical Engineering and Computer Science **June 2005**
- Advisor: Professor Michael D. Ernst

**University of Utah**, Salt Lake City, Utah, USA

B.S., Computer Science (with Honors) **May 2003**
- Advisor: Professor Wilson Hsieh

B.S., Mathematics **May 2003**

AWARDS AND FELLOWSHIPS

**Alfred P. Sloan Research Fellowship**, 2015

**ASHG Trainee Research Semifinalist Award**, 2011

**Ruth L. Kirschstein National Research Service Award (NRSA)**, 2010
NIH post-doctoral fellowship (2010 – 2013)

**NSF Graduate Research Fellowship**, 2003
Pre-doctoral fellowship (2003 – 2006)

**Dean's List**, University of Utah, 1998 – 2002

**C.M. Collins Engineering Scholarship**, University of Utah, 2002

**Early undergraduate admission** following high school junior year, 1998

PROFESSIONAL EXPERIENCE

**23andMe**, Sunnyvale, California, USA

*Senior Scientist II* **January 2022 to November 2024**
Population Genetics Research & Development

**Cornell University**, Ithaca, New York, USA

*Associate Professor* **July 2020 to December 2022**
*Nancy and Peter Meinig Family Investigator in Life Sciences*

*Assistant Professor* **August 2014 to June 2020**

**Columbia University**, New York, New York, USA, *and*
**Howard Hughes Medical Institute**, Chicago, Illinois, USA
*Work conducted at* ***Columbia University***

*Postdoctoral Research Fellow* **August 2013 to July 2014**
- Advisor: Professor Molly Przeworski

**Harvard Medical School**, Boston, Massachusetts, USA

*Postdoctoral Research Fellow* **October 2009 to July 2013**
- Advisors: Professors David Reich and David Altshuler

**Massachusetts Institute of Technology**, Cambridge, Massachusetts, USA

*Graduate Student and Research Assistant*      **January 2004 to October 2009**
- Advisors: Professors David K. Gifford and David E. Housman

*Teaching Assistant*      **January 2005 to May 2005**
- 6.170, Spring Semester 2005: Laboratory in Software Engineering
  – Undergraduate course in software engineering principles

**University of Utah**, Salt Lake City, Utah, USA

*Undergraduate Research Assistant*      **May 2002 to May 2003**
- Advisors: Professors Wilson Hsieh and Peter Shirley

PREPRINTS      Qiao Y, Jewett EM, McManus KF, Freyman WA, Curran JE, Williams-Blangero S, Blangero J, The 23andMe Research Team, **Williams AL**[#]. *Reconstructing parent genomes using siblings and other relatives.* (**bioRxiv preprint**) doi:10.1101/2024.05.10.593578.

Williams CM[#], O'Connell J, Freyman WA, 23andMe Research Team, Gignoux CR, Ramachandran S[#], **Williams AL**[#]. *Phasing millions of samples achieves near perfect accuracy, enabling parent-of-origin classification of variants.* (**bioRxiv preprint**) doi:10.1101/2024.05.06.592816.

Avadhanam S, **Williams AL**[#]. *Phase-free local ancestry inference mitigates the impact of switch errors on phase-based methods.* (**bioRxiv preprint**) doi:10.1101/2023.12.02.569669.

PUBLICATIONS      [#]Corresponding author   [*]Co-first author   Underlined: student/postdoctoral advisee

Massarat AR, Lamkin M, Reeve C, **Williams AL**, D'Antonio M, Gymrek M. *Haptools: a toolkit for admixture and haplotype analysis.* **Bioinformatics 39(3):btad104**, Mar. 2023.

Avadhanam S, **Williams AL**[#]. *Simultaneous inference of parental admixture proportions and admixture times from unphased local ancestry calls.* **Am J Hum Genet 109(1):1405-1420**, Aug. 2022.

Smith J[*], Qiao Y[*,#], **Williams AL**[#]. *Evaluating the utility of identity-by-descent segment numbers for relatedness inference via information theory and classification.* **G3 Genes|Genomes|Genetics**, Mar. 2022.

Chan AW, Villwock SS, **Williams AL**, Jannink JL. *Sexual dimorphism and the effect of wild introgressions on recombination in Manihot esculenta.* **G3 Genes|Genomes| Genetics 12(1):jkab372**, Nov. 2021.

Kivisild T, Saag L, Hui R, Biagini SA, Pankratov V, D'Atanasio E, Pagani L, Saag L, Rootsi S, Mägi R, Metspalu E, Valk H, Malve M, Irdt K, Reisberg T, Solnik A, Scheib CL, Seidman DN, **Williams AL**, Tambets K, Metspalu M. *Patterns of genetic connectedness between modern and medieval Estonian genomes reveal the origins of a major ancestry component of the Finnish population.* **Am J Hum Genet 108(9):1792-1806**, Sep. 2021.

Qiao Y[*], Sannerud J[*], Basu-Roy S, Hayward C, **Williams AL**[#]. *Distinguishing pedigree relationships using multi-way identity by descent sharing and sex-specific genetic maps.* **Am J Hum Genet 108(1):68-83**, Jan. 2021.

<u>Hubisz MJ</u>, **Williams AL**, Siepel, A. *Mapping gene flow between ancient hominins through demography-aware inference of the ancestral recombination graph.* **PLOS Genet 16(8):e1008895**, Aug 2020.

<u>Seidman DN</u>, Shenoy SA, <u>Kim M</u>, <u>Babu R</u>, Dyer TD, Lehman DM, Curran JE, Duggirala R, Blangero J, **Williams AL**[#]. *Rapid, phase-free detection of long identity-by-descent segments enables effective relationship classification.* **Am J Hum Genet 106(4):453-466**, Apr. 2020.

<u>Caballero M</u>, <u>Seidman DN</u>, <u>Qiao Y</u>, <u>Sannerud J</u>, Dyer TD, Lehman DM, Curran JE, Duggirala R, Blangero J, Carmi S, **Williams AL**[#]. *Crossover interference and sex-specific genetic maps shape identical by descent sharing in close relatives.* **PLOS Genet 15(12):e1007979**, Dec 2019.

Chan AW, **Williams AL**, Jannink JL. *A statistical framework for detecting mislabeled and contaminated samples using shallow-depth sequence data.* **BMC Bioinformatics 19:478**, Dec 2018.

<u>Ramstetter MD</u>, Dyer TD, Lehman DM, Curran JE, Duggirala R, Blangero J, Mezey JG, **Williams AL**[#]. *Inferring identical by descent sharing of sample ancestors promotes high resolution relative detection.* **Am J Hum Genet 103(1):30-44**, Jul 2018.
• Video of invited webinar presentation from Broad Institute [YouTube]

Mercader JM, Liao RG, Bell AD, Dymek Z, Estrada K, Tukianinen T, … [18 authors] … **Williams AL**, … [54 authors] … Altshuler D, Florez JC on behalf of the SIGMA T2D Genetics Consortium. *A loss-of-function splice acceptor variant in IGF2 is protective for type 2 diabetes.* **Diabetes 66(11):2903-2914**, Nov 2017.

<u>Ramstetter MD</u>[#], Dyer TD, Lehman DM, Curran JE, Duggirala R, Blangero J, Mezey JG, **Williams AL**[#]. *Benchmarking relatedness inference methods with genome-wide data from thousands of relatives.* **Genetics 207(1):75-82**, Sep 2017.

Palmer ND, Goodarzi MO, Langefeld CD, … [22 authors] … **Williams AL**, … [21 authors] … Watanabe RM, Wagenknecht LE. *Genetic Variants Associated with Quantitative Glucose Homeostasis Traits Translate to Type 2 Diabetes in Mexican Americans: The GUARDIAN (Genetics Underlying Diabetes in Hispanics) Consortium.* **Diabetes 64(5):1853-66**, May 2015.

**Williams AL**[#], Genovese G, Dyer T, et al. for the T2D-GENES Consortium. *Non-crossover gene conversions show strong GC bias and unexpected clustering in humans.* **eLife 2015;4:e04637**, Apr 2015.

The SIGMA Type 2 Diabetes Consortium: Estrada K, Aukrust I, Bjørkhaug L, Burtt NP, Mercader JM, García-Ortiz H, Huerta-Chagoya A, Moreno-Macías H, Walford G, Flannick J, **Williams AL**[†], et al. *Association of a Low-Frequency Variant in HNF1A With Type 2 Diabetes in a Latino Population.* **JAMA 311(22):2305-14**, Jun 2014.
[†]**Performed sample selection for exome sequencing and contributed to association analyses**

The SIGMA Type 2 Diabetes Consortium: **Williams AL**[‡], Jacobs SBR, Moreno-Macías H, et al. *Sequence variants in SLC16A11 are a common risk factor for type 2 diabetes in Mexico.* **Nature 506:97-101**, Feb 2014.
[‡]**Led the genetics analysis and wrote the genetics portions of paper.**

The 1000 Genomes Project Consortium[§]. *An integrated map of genetic variation from 1,092 human genomes.* **Nature 491:56-65**, Nov 2012.

[§]**Developed HAPMIX extension and applied it to the Latino populations.**

**Williams AL**[#], Patterson N, Glessner J, Hakonarson H, Reich D. *Phasing Many Thousands of Genotyped Samples.* **Am J Hum Genet, 91:238-51**, Aug 2012.

Fejerman L, Chen GK, Eng C, Huntsman S, Hu D, **Williams A**, Pasaniuc B, John EM, Via M, Gignoux C, Ingles S, Monroe KR, Kolonel LN, Torres-Mejía G, Pérez-Stable EJ, Burchard EG, Henderson BE, Haiman CA, Ziv E. *Admixture mapping identifies a locus on 6q25 associated with breast cancer risk in US Latinas.* **Hum Mol Genet, 21:1907-17**, Apr 2012.

**Williams AL**[#], Housman DE, Rinard MC, Gifford DK. *Rapid Haplotype Inference for Nuclear Families.* **Genome Biol, 11:R108**, Oct 2010.

**Williams A**, Thies W, Ernst MD. *Static Deadlock Detection for Java Libraries.* **In Proceedings of the 2005 European Conference on Object-Oriented Programming (ECOOP)**. Glasgow, Scotland, UK. Jul 2005.

**Williams A**, Barrus S, Morley K, Shirley P. *An efficient and robust ray-box intersection algorithm.* **Journal of Graphics Tools, 10:49-54**, Jun 2005.

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| GRANT SUPPORT | NIH/NIGMS R35GM133805 (Role: **PI**) |

Scalable methods for the characterization and analysis of families in large genomic datasets.

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| Award period: | 9/1/2019 – 7/31/2024 |
| Total amount: | $1,606,482 |

Alfred P. Sloan Research Fellowship, Alfred P. Sloan Foundation (Role: **PI**)

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| Award period: | 9/15/2015 – 9/14/2017 |
| Total amount: | $50,000 |

NIH/NIGMS R01GM102192 (Role: **Subaward PI**; PI: Adam Siepel)

Computational methods for human genomic data integration: demography, selection, and functional potential.

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|---|---|
| Award period | 9/1/2014 – 8/31/2017 |
| Total amount: | $148,441 |

CONFERENCE TALKS

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|---|---|
| *"Reconstructing parent genotypes at genotyping array accuracy using siblings and other relatives"* | |
| Society for Molecular Biology and Evolution | July 2024 |

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| *"Estimating relationships by combining DNA from multiple siblings"* | |
| RootsTech | March 2022 |

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| *"Exploiting multi-way identity-by-descent to detect pedigree relationships, cluster relatives, and infer long-range phase"* (**Invited Speaker**) | |
| Society for Molecular Biology and Evolution | July 2021 |

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| *"Reconstructing parent DNA and evaluating relatives at HAPI-DNA.org"* | |
| RootsTech | February 2021 |

*"Reconstructing Grandma's Genome"*
RootsTech                                                            February 2020

*"Using DNA from many samples to distinguish pedigree relationships of close relatives"*
Family History Technology Workshop                                   February 2020

*"Inferring identical by descent sharing of sample ancestors promotes high resolution relatedness detection"*
Probabilistic Modeling in Genomics (PROBGEN)                         September 2017

*"Inferring the genomes of mothers and fathers using genotype data from a set of siblings"*
Family History Technology Workshop                                   February 2017

*"Pedigree reconstruction in the era of many thousands of samples"*
Probabilistic Modeling in Genomics (PROBGEN)                         September 2016

*"Inferring local ancestry by jointly analyzing admixed samples"* **(Invited Speaker)**
Probabilistic Modeling in Genomics (PROBGEN)                         October 2015

*"Inferring local ancestry by jointly analyzing admixed samples"* **(Invited Speaker)**
Models and Inference in Population Genetics Workshop                 September 2015

*"Fine-scale properties of non-crossover gene conversion in humans."* **(Invited Speaker)**
New York Area Population Genomics Workshop                           January 2015

*"The fine-scale landscape of meiotic non-crossover gene conversion."*
64th Annual Meeting of The American Society of Human Genetics        October 2014

*"Non-crossover gene conversions show strong allelic bias and unexpected clustering in humans"*
Biological Sequence Analysis and Probabilistic Models Workshop       July 2014

*"A genome-wide estimate of the meiotic gene conversion rate in humans."*
Biology of Genomes                                                   May 2013

*"Identification of a novel genome-wide significant association with type 2 diabetes risk in Mexican and Mexican Americans."*
62nd Annual Meeting of The American Society of Human Genetics        November 2012

*"Phasing of Many Thousands of Genotyped Samples."*
61nd Annual Meeting of The American Society of Human Genetics        October 2011
**Semifinalist for Trainee Research Award**

*"Static Deadlock Detection for Java Libraries."*
European Conference on Object-Oriented Programming (ECOOP)           July 2005

INVITED TALKS   *"Reconstructing parental genomes and cross-chromosome phasing using data from millions of people"*
                – UC Berkeley                                        November 2024
                – Brigham Young University                           November 2024

*"Reconstructing parental genomes and near perfect phasing using data from millions of people"*

  – Stanford University                                                       May 2024
  – Women in Data Science (Stanford University)            May 2024
  – University of Georgia                            March 2024

*"Inferring haplotypes with millions of samples and estimating parent admixture proportions and times"*

  – Brown University                  January 2023

*"Reconstructing parent genotypes using data from siblings"*

  – New York Genome Center           April 2022

*"New methods to classify pedigree relationships and reconstruct parent DNA"*

  – University of Southern California       February 2022

*"Exploiting multi-way identity-by-descent to detect pedigree relationships, cluster relatives, and infer long-range phase"*

  – University of Utah                 October 2021

*"New methods for classifying relatives in large datasets"*

  – 23andMe                        July 2020

*"Distinguishing pedigree relationships using multi-way identity by descent segments detected in phase-free genotypes"*

  – Mount Sinai                   November 2019

*"Reconstructing ancestor genomes using genetic data from close relatives"*

  – FamilySearch                  April 2019
  – Utah Valley University           April 2019

*"Inferring identical by descent sharing of sample ancestors promotes high resolution relative detection"*

  – Broad Institute of Harvard and MIT (Web-based talk)      May 2018

*"Leveraging and inferring properties of ungenotyped ancestors enables high resolution relatedness detection"*

  – Brigham Young University          April 2018
  – 23andMe                        April 2018
  – Ithaca College                February 2018
  – Brown University            November 2017

*"Relatedness inference in modern samples: Leveraging multi-way signals to improve resolution"*

  – Weill Cornell Medicine           March 2017

*"Towards large-scale disease and ancestry analyses in diverse samples"*

  – Population Architecture using Genomics and Epidemiology (PAGE)
     Steering Committee Meeting          November 2015

*"Haplotype inference of large datasets and applications to gene conversion and disease studies"*

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| – Brigham Young University | March 2014 |
| – Cornell University | February 2014 |
| – New York University | February 2014 |
| – University of Chicago | February 2014 |

*"Meiotic gene conversion in humans: rate, sex ratio, and GC bias"*

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| – University of Chicago | June 2013 |

*"Analyzing gene conversion event rate and structure"*

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| – Texas Biomedical Research Institute | April 2013 |

*"Sequence variants in SLC16A11 are a common risk factor for type 2 diabetes in Mexico"*

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| – Icahn School of Medicine at Mount Sinai | November 2013 |
| – Texas Biomedical Research Institute | April 2013 |
| – Harvard Medical School | March 2013 |
| – Broad Institute Annual Retreat | November 2012 |

*"Phasing of Many Thousands of Genotyped Samples"*

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| – Harvard Medical School | March 2013 |
| – Columbia University | February 2013 |
| – Broad Institute of Harvard and MIT | February 2013 |

OUTREACH

Informing the broader non-scientific community about my research is a passion I spend both time and a small amount of personal money to pursue. I am especially interested in reaching the lay genetic genealogy community, an avid group of family history practitioners. To that end, I have presented at the RootsTech family history conference three times, with recorded videos from some pandemic years available on YouTube. I also built and continue to maintain the website https://hapi-dna.org/, which provides user-friendly web-based versions of several tools my lab developed. (Notably I built the web versions of the tools and did not ask trainees to do so.) The website also includes helpful information such as the rates at which various relatives share identity-by-descent (IBD) segments. It is linked to from the International Society of Genetic Genealogy (ISOGG) website and receives an average of roughly 50 views per day.

PROFESSIONAL SERVICE

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| Ad hoc reviewer, National Institutes of Health (MRAA) Study Section | 2024 |
| Reviewer, National Science Foundation (CISE) Panel | 2019 |
| Ad hoc reviewer, National Institutes of Health (BMRD) Study Section | 2018 |
| Reviewer, Intelligent Systems for Molecular Biology (ISMB)/ECCB Conference | 2018 |
| Reviewer, National Heart Lung and Blood Institute (NHLBI) Special Emphasis Panel | 2016 |
| Reviewer, National Human Genome Research Institute (NHGRI) Special Emphasis Panel | 2015 |
| Reviewer, American Society of Human Genetics 65th Annual Meeting | 2015 |
| Reviewer, Intelligent Systems for Molecular Biology (ISMB)/ECCB Conference | 2015 |
| Ad hoc reviewer for manuscripts submitted to the following journals: | 2010-Present |

◇Guest editor
– Nature
– Nature Genetics
– eLife◇
– Proceedings of the National Academy of Sciences

– Nature Communications
– Genome Research
– American Journal of Human Genetics
– Molecular Biology and Evolution
– PLOS Genetics$^\diamond$
– PLOS Computational Biology
– Bioinformatics
– Genetics
– European Journal of Human Genetics
– Annals of Human Genetics
– Diabetologia
– PLOS ONE
– Molecular Ecology Resources

| UNIVERSITY SERVICE | The following service was performed at Cornell University: | |
|---|---|---|
| | – Director of Graduate Studies, Computational Biology | 2020-2021 |
| | – Faculty Search Committee, Computational Biology | 2019-2021 |
| | – Admissions Committee, Computational Biology and/or Computational Biology and Medicine Graduate Fields | 2014-2021 |
| | – Lead PI overseeing Computational Biology Compute Cluster | 2018-2021 |
| | – Advisor to up to 13 undergraduate students majoring in Biometry & Statistics and/or Biological Sciences | 2015-Present |
| | – Advisor to up to 10 freshman Biological Sciences majors | 2017-2019 |
| | – Faculty Search Committee, Computational Biology | 2018-2019 |
| | – Senator, CALS Faculty Senate | 2017-2018 |
| | – Program Committee, Computational Biology and Medicine Graduate Field Tri-Institutional program of Cornell University, Weill Cornell Medical College, and Sloan-Kettering Institute | 2016-2017 |
| | – Faculty Search Committee, Biological Statistics & Computational Biology | 2015-2016 |
| | – Consulted Life Science Advisory Committee concerning computational biology on campus | Oct 2015 |
| | – Faculty Search Committee, Molecular Biology and Genetics Dept. | 2014-2015 |

TEACHING     Computational Genetics and Genomics: BTRY 4840/6840, cross-listed as CS 4775 (Fall semester, every year from 2015 to 2020), Cornell University. Up to 57 undergraduate and graduate students. Introduction to computational algorithms for analyzing genetic and genomic data. Assignments include bi-weekly statistical and Python-based computational problem sets and a final project implementing an algorithm from the literature or a new method typically based on the student's research.

Statistical and Computational Genetics: BTRY 7200 (Spring semester, every year from 2016 to 2021), Cornell University. 10 students, primarily graduate. Weekly readings and discussions of recent literature on computational genetics or population genetics.

MENTORING    **Doctoral students**
Siddharth Avadhanam, Computational Biology, Cornell University, 2018-2023.
Jens Sannerud, Genetics, Genomics, & Development, Cornell University, 2017-2023.
Daniel Seidman, Computational Biology & Medicine, Cornell University, 2017-2022.
Ying Qiao, Computational Biology, Cornell University, 2017-2021.
Melissa Hubisz, Computational Biology, Cornell University, co-chair, 2014-2019.
Monica Ramstetter, Computational Biology, Cornell University, co-chair, 2015-2017.

**Doctoral committees**
Cole M. Williams, Computational Biology, Brown University, 2021-Present.
Travis Wrightsman, Plant Breeding, Cornell University, 2019-2024.
Runxi Shen, Computational Biology, Cornell University, 2018-2022.
Jonathan Hughes, Ecology & Evolutionary Biology, Cornell University, 2017-2022.
Afrah Shafquat, Computational Biology, Cornell University, 2017-2021.
Konnor La, Computational Biology & Medicine, Cornell University, 2020.
Ariel Chan, Plant Breeding, Cornell University, 2016-2019.

**Master's students**
Rajesh Bollapragada, M.Eng., Computer Science, 2019.
Ramya Babu, M.S., Computer Science, 2017-2018.
Joshua Cohen, M.Eng., Computer Science, 2016.

**Undergraduate researchers**
Yilei Huang, B.S., Biometry & Statistics, 2020-2021.
Michael Merrill, B.S., Information Science, 2017.
Arjun Biddanda, B.S., Computer Science, 2015.

*Updated November 18, 2024*