Amy L. Williams

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 Present**
Product 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

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.

Hubisz MJ, **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.

Seidman DN, Shenoy SA, Kim M, Babu R, 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.

Caballero M, Seidman DN, Qiao Y, Sannerud J, 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.

Ramstetter MD, 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.

Ramstetter MD[#], 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.

[†]**Preformed 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.

Grant Support

NIH/NIGMS R35GM133805 (Role: **PI**)
Scalable methods for the characterization and analysis of families in large genomic datasets.
Award period: 9/1/2019 – 7/31/2024
Total amount: $1,606,482

Alfred P. Sloan Research Fellowship, Alfred P. Sloan Foundation (Role: **PI**)
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.
Award period 9/1/2014 – 8/31/2017
Total amount: $148,441

Conference Talks

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

"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

"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

"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"

– 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"

– University of Chicago June 2013

"Analyzing gene conversion event rate and structure"

– Texas Biomedical Research Institute April 2013

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

– 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"

– Harvard Medical School March 2013

– Columbia University February 2013
– Broad Institute of Harvard and MIT February 2013

Professional Service

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

$^{\diamond}$Guest editor
– Nature
– Nature Genetics
– eLife$^{\diamond}$
– 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

Cornell University Service

– 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 7 Oct 2015
– Faculty Search Committee, Molecular Biology and Genetics Dept. 2014-2015

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