#### Andrew R. Marderstein

Postdoctoral Fellow | Stanford University

Mobile: (914) 806-1890 | E-Mail: amarder@stanford.edu Twitter: @amarderstein | GitHub: github.com/drewmard

## **Education**

## Postdoctoral Fellow, Stanford University

2021 - 2024

School of Medicine Dean's Fellow in the Department of Pathology.

PI: Stephen Montgomery. Close collaborators: Ana Cvejic (Cambridge/Copenhagen), Anshul Kundaje (Stanford).

## PhD - Computational Biology & Medicine (CBM), Tri-Institutional Program

2017 - 2021

Cornell University / Weill Cornell Medical College | Memorial Sloan Kettering Cancer Center | Rockefeller University Pls: Andrew Clark (CU) and Olivier Elemento (WCMC).

## **BS**, Cornell University

2013 - 2017

Major in Biometry & Statistics (Concentration: Statistical Genomics) and a Minor in Biological Sciences. PI: Philipp Messer. Graduated with honors and distinction in research. Departmental (Major) GPA: 4.00.

## **Research Experience**

#### **Memorial Sloan Kettering Cancer Center**

2024 - Present

Research Fellow, Division of Clinical Genetics (with Dr. Kenneth Offit)

Studying high-risk cancer patients

#### **Stanford University**

2021 - 2024

Postdoctoral Fellow, Dr. Stephen Montgomery Lab

• Combining human genetics and single-cell multi-omics to identify disease mechanisms

## Third Rock Ventures

2022 - 2023

External Consultant at Marea Therapeutics

Discovering therapeutic targets using RNA-seq + GWAS

**Pfizer**2021 – 2022
External Consultant (with Dr. Randy Longman at Weill Cornell Medicine), Biomarker & Precision Medicine Team

Modeling of multi-omic data from clinical trials to predict patient response and disease progression

## Cornell University and Weill Cornell Medical College

2017 - 2021

PhD Candidate; Dr. Andrew Clark Lab and Dr. Olivier Elemento Lab

Thesis: Analyzing modifiable risk factors in disease using human genetic variation (e.g. GxE interactions)

#### **Regeneron Genetics Center**

Summer 2016/Summer 2017

Statistical Genetics Intern; Supervised by Dr. Cristopher Van Hout and Dr. Shane McCarthy

• Evaluated the false positive rate and power of statistical methods commonly-used during GWAS

## Cornell Department of Biological Statistics and Computational Biology

2015 - 2017

Undergraduate Honors Researcher; Dr. Philipp Messer Lab

Developed a Bayesian coalescent framework for studying selective sweeps in population genetic data

#### **Cornell Department of Natural Resources**

2016 - 2017

Statistician - Consultant for Hudson River Fisheries Unit; Supervised by Dr. Pat Sullivan

Created quantitative models of fish population dynamics in time-series data

#### **Cornell Institute for Resource Information Sciences (IRIS)**

2015 - 2016

Undergraduate Researcher; Supervised by Dr. Magdeline Laba

• Developed New York State 4-H youth education programs; mapped invasive species in the Hudson River

## **Presented Work**

**Publications:** (^ or \* = co-first or co-corresponding authors)

### Under Review/In Revision:

- **1.** IGVF Consortium (2024). <u>The Impact of Genomic Variation on Function (IGVF) Consortium</u>. Preprint on *arXiv*. doi: https://doi.org/10.48550/arXiv.2307.13708 (Under review at *Nature*)
- 2. Rubenstein, A.B., Smith, G.R., Zhang, Z., ..., A.R. Marderstein, S.B. Montgomery, ..., Trappe, S.\*, Sealfon, S.C.\*. Integrated single-cell multiome analysis reveals muscle fiber-type gene regulatory circuitry modulated by endurance exercise. Preprint on *bioRxiv*. doi: 10.1101/2023.09.26.558914 (Under review at *Cell Reports Medicine*)
- 3. Kulm, S., Marderstein, A.R., Mezey, J.\*, Elemento, O.\* <u>A systematic framework for assessing the clinical impact of polygenic risk scores</u>. Preprint on *medRxiv*. doi: https://doi.org/10.1101/2020.04.06.20055574 (In revision)
- **4.** Krishna, C., Walker, R.L., **Marderstein, A.R.**, Brown, E.M., Deguine, J., Graham, D.B., Xavier, R.J. <u>Immunogenetic control of gene expression and cell state</u>. (In revision)
- 5. Kong, Y., Harrington, D., Marderstein, A.R., Alonso, L.C. <u>Low frequency T2D-associated polymorphisms at CCND2 influence CCND2 mRNA abundance, but not proliferation, in human pancreatic islets</u>. (In revision)

## Published/Accepted:

- **1. Marderstein, A.R.**, De Zuani, M., Moeller, R., Bezney, J., Padhi, E., Wong, S., Coorens, T.H.H., Xie. Y., Xue, H., Montgomery, S.B.\*, Cvejic, A\* (2024). <u>Single-cell multi-omics map of human foetal blood in Down's Syndrome</u>. Preprint on *bioRxiv*. doi: https://doi.org/10.1101/2023.09.25.559431 (Accepted at *Nature*)
- Lima, S., Rupert, A., Putzel, G., Marderstein, A.R., Woo, V., Viladomiu, M., Metz, M., Scherl, E., Longman, R (2024). The gut microbiome regulates the clinical efficacy of sulfasalazine therapy for IBD-associated spondyloarthritis. Cell Reports Medicine. doi: https://doi.org/10.1016/j.xcrm.2024.101431.
- 3. Shah, Y.^, Verma, A.^, Marderstein, A.R., White, J., Bhinder, B., Medica, J.S.G., Elemento, O (2021). <u>Pan-cancer analysis reveals molecular patterns associated with age.</u> *Cell Reports* 37, 110100. doi: https://doi.org/10.1016/j.celrep.2021.110100.
  - > Precision Oncology News: "Pan-Cancer Analysis Reveals Unique Features in Tumors From Younger People"
- **4.** Marderstein, A.R., Kulm, S., Peng, C., Tamimi, R.M., Clark, A.G.\*, Elemento, O.\* (2021). <u>A polygenic score-based approach for identification of gene-drug interactions stratifying breast cancer risk</u>. *The American Journal of Human Genetics* 108 (9), 1752-1764. doi: 10.1016/j.ajhg.2021.07.008.
  - > GenomeWeb: "Breast Cancer Genetic Risk Modified by Corticosteroid Use"
- 5. **Marderstein, A.R.**, Davenport, E.R., Van Hout, C.V., Kulm, S., Elemento, O.\*, Clark, A.G.\* (2021). <u>Leveraging phenotypic variability to identify genetic interactions in human phenotypes</u>. *American Journal of Human Genetics* 108 (1), 1-19. https://doi.org/10.1016/j.ajhg.2020.11.016
  - > Cornell Chronicle: "Cross-campus team probes gene-environment interactions"
  - > Weill Cornell Newsroom: "Researchers simplify the study of gene-environment interactions"
- **6. Marderstein, A.R.**, Uppal, M., Verma, A., Bhinder, B., Tayyebi, Z., Mezey, J., Clark, A.G.\*, Elemento, O.\* (2020). <u>Demographic and genetic factors influence the abundance of infiltrating immune cells in human <u>tissues</u>. *Nature Communications* 11, 2213. https://doi.org/10.1038/s41467-020-16097-9</u>
- 7. Castellanos, J.G., Woo, V., Viladomiu, M., Putzel, G., Lima, S., Diehl, G.E., Marderstein, A.R., Gandara, J., Perez, A.R., Withers, D.R., Targan, S.R., Shih, D.Q., Scherl, E.J., Longman, R.S. (2018). Microbiota-induced TNF-like ligand 1A drives Group 3 innate lymphoid cell-mediated barrier protection and intestinal T cell activation during colitis. *Immunity*. 49(6):1077-1089.e5. doi: 10.1016/j.immuni.2018.10.014
- 8. Marderstein, A.R. (2017). <u>Approximate Bayesian Computation for Studying Selective Sweep Signatures in Local Coalescence Trees</u>. Honors Thesis, College of Agriculture and Life Sciences, Cornell University. Supervisor: Philipp Messer.

# **Platform Presentations:**

Conference:						
[Presenter]						
2024	American Society of Human Genetics conference in Denver, CO (Platform title: "Genetics of Human Brain: Regulation, Disease Risk, and Assortative Mating")					
2023	American Society of Human Genetics conference in Washington D.C.					
	(Platform title: "Causal non-coding variants and the genes they impact")					
2023	IGVF Annual Meeting in St. Louis, MO					
2022	American Society of Human Genetics conference in Los Angeles, CA (Platform title: "Genetic Impacts on the Epigenome and Beyond")					
	> Semifinalist: 2022 Charles J. Epstein Trainee Awards for Excellence in Human Genetics Research					
2022	dGTEx Annual Meeting in Bethesda, MD (on behalf of the Data Analysis & Assays WG)					
2022	IGVF Annual Meeting in Bethesda, MD (on behalf of the Immune Disease FG)					
2020	eKeystone Virtual Symposia: Beyond a Million Genomes ( <i>Invited Talk</i> ) (Virtual)					
2020	Beyond a Million Genomes: From Discovery to Precision Health, Keystone Symposia, Breckenridge, CO.					
2019	American Society of Human Genetics conference in Houston, TX.					
	(Platform title: "Mechanisms of Immune Cell Phenotypes and Clonal Hematopoiesis")  > Semifinalist: 2019 Charles J. Epstein Trainee Awards for Excellence in Human Genetics Research					
2019	New York Academy of Sciences: Translating Genetics into Medicine Conference in New York, NY.					
2018	Population, Evolutionary, and Quantitative Genetics conference in Madison, WI.					
[Senior author]						
2022	American Society of Human Genetics conference in Los Angeles, CA (led by Lin Poyraz)					
	(Platform title: "The current environment for gene-environment interactions")					
2021	Society for Molecular Biology & Evolution (led by Lin Poyraz)					
	(Platform title: "Genotype-environment interactions in the genomics era")					
<u>Internal:</u>						
2024	University of Chicago, Chicago, IL					
2024	Biotech Research Innovation Centre (BRIC), University of Copenhagen, Denmark (Invited Talk)					
2024	Case Western Reserve University, Columbia, OH					
2024	Inst. of Stem Cell Biology & Regenerative Medicine at Stanford University, Stanford, CA					
2024	Memorial Sloan Kettering Cancer Center, New York, NY					
2024	Columbia University, New York, NY					
2023	Yale University, New Haven, CT (Invited Talk)					
2023	Center for Human Genetics & Genomics, NYU, New York, NY					
2023	Division of Precision Medicine, Department of Medicine, NYU, New York, NY					
2023	IGVF: Non-coding Variants FG (Virtual), IGVF Immune Disease FG (Virtual)					
2023	Pediatric Computational Biology Group at Stanford University, Stanford, CA					
2022	IGVF: Computation, Analysis, Modeling, Prediction WG (Virtual), Phenotypic Impact FG (Virtual),					
	Immune Disease FG (Virtual)					
2021	Tri-I Computational Biology & Medicine Seminar at Weill Cornell Medicine (Virtual)					
2021	Student Talk at Tri-I Computational Biology & Medicine Program Recruitment (Virtual)					
2021	Department of Computational Medicine, UCLA (Invited Talk) (Virtual)					
2020	Analytical & Translational Genetics Unit, Broad Institute (Invited Talk) (Virtual)					
2020	Student Talk at Tri-I Computational Biology & Medicine Retreat (Virtual)					
2020	Tri-I Computational Biology & Medicine Seminar at Weill Cornell Medicine, New York, NY					
2020	Computational Biology Student Seminar at Cornell University, Ithaca, NY					
2019	Basic Research Working Group meeting at Weill Cornell Medicine, New York, NY					
2019	Panelist, Precision Medicine Symposium at Weill Cornell Medicine, New York, NY					
2010	(Panel discussion title: "Facilitating Cross Campus Collaboration for Trainees")					
2019 2019	Microbiome Meeting at Cornell University, Ithaca, NY Invited alumni papeliet, Cornell Accepted Students Reconting in Rya, NY					
	Invited alumni panelist, Cornell Accepted Students Reception in Rye, NY					
2019 2017	Computational Biology Student Seminar at Cornell University, Ithaca, NY					
2017	Regeneron Genetics Center in Tarrytown, NY Intern Bl-B. Regeneron Pharmacouticals. Inc in Tarrytown, NY					
2010	Intern B&B, Regeneron Pharmaceuticals, Inc in Tarrytown, NY  > Intern project was selected to be part of a company-wide telecast and presentation					
	Intern project was selected to be part of a company-wide telecast and presentation					

#### **Poster Presentations:**

<u>Conference:</u>					
2024	IGVF Annual Meeting in Seattle, WA				
2023	IGVF Annual Meeting in St. Louis, MO				
2023	Biology of Genomes conference in Cold Spring Harbor Laboratory, NY				
2021	American Society of Human Genetics conference (held virtually due to the COVID-19 pandemic)				
2020	American Society of Human Genetics conference (held virtually due to the COVID-19 pandemic)				
	> Awarded "Reviewer's Choice" for top 10% of all submitted abstracts.				
2020	Beyond a Million Genomes: From Discovery to Precision Health, Keystone Symposia in Breckenridge, CO				
2019	New York Academy of Sciences: Translating Genetics into Medicine Conference in New York, NY				
	> Awarded the "F1000 Outstanding Presentation Prize" for best poster.				
2018	American Society of Human Genetics conference in San Diego, CA				
2018	Population, Evolutionary, and Quantitative Genetics conference in Madison, WI				
Local:					
2020	Tri-I Symposium in Computational Biology & Medicine at Memorial Sloan Kettering Cancer Center, New York, NY				
2019	39th Annual Vincent du Vigneaud Student Research Symposium at Weill Cornell Medicine, New York, NY				
2017	Honors Symposium, Cornell University in Ithaca, NY				
2016	Cornell Undergraduate Research Board Fall Forum in Ithaca, NY				

## **Leadership and Service**

#### **Current:**

Co-Chair of the Career Development Committee, American Society of Human Genetics

2020 - 2024

- Organize, moderate, and lead career fairs, panels, workshops, and resources for early-career members, such as:
- Host of the "Trainee Paper Spotlight" podcast, bringing stories from up-and-coming scientists into the spotlight
- Lead organizer of the "Entrepreneurship in Human Genetics" panel at the 2020 meeting
- Co-organizer and panelist for the "Tips on How to Land a Postdoctoral Position" at the 2021 meeting
- Founded the "Trainee Social Reception" at the 2022 meeting; now a yearly event (>400 attendees)
- Co-organizer of "The Confidence Factor: Thriving in Your Career" at the 2023 meeting
- Co-organized a bimonthly "Work-Life Balance" webinar series on rotating themes from 2021-2023

2023 IGVF Immune Trainee Focus Group Co-Lead, IGVF

2023 - Present

2023 IGVF All-Consortium Monthly Meeting Organizing Committee, IGVF

2023 - Present

#### Speaker, Skype a Scientist

2017 - Present

• Chat about fascinating genetics research with 7th to 12th grade classrooms, with an emphasis on captivating students' interests and clearly explaining complex topics to students that may have limited scientific backgrounds. I've engaged over 1,000 students total around the country through this program.

#### Fundraiser, Crohn's & Colitis Foundation of America (CCFA)

2011 - Present

• Fundraising to promote awareness and find a cure. Half-marathon Team Challenge finisher.

#### **Previous:**

2023 IGVF Annual Meeting Planning Committee, Impact of Genomic Variation on Function (IGVF)

2023

• In addition to helping organize the annual meeting for 200+ attendees, I moderated a session and led a break-out discussion during the meeting.

#### 2023 Annual Meeting Abstract Reviewer, American Society of Human Genetics

2023

Section: Complex Traits and Polygenic Disorders III: Infectious disease and immunological disorders

Focus Group for evaluating a professional Certification Program, American Society of Human Genetics

2023

2022 Meeting Planning Advisory Group, American Society of Human Genetics 2022 Provide counsel to the staff in the face of opportunities, challenges, and costs of a return in a new environment E.g. how to communicate vaccination requirements, or how to deliver hybrid (remote/in-person) content Search Committee for Diversity, Inclusion and Belonging Position, Stanford Genetics Department 2022 Postdoctoral Representative of the committee helping build a diverse and inclusive community at Stanford Judge, Amgen Scholars - Stanford Summer Research Program (SSRP) Symposium 2022 2022 Lead Organizer, Montgomery Lab Retreat in Lake Tahoe, CA Guest Lecturer, BIOS 2000: "Foundations in Experimental Biology", Stanford University 2021 President, Tri-Institutional Biotech Club 2019 - 2021 The largest club on campus (includes students and postdocs across Cornell, Sloan Kettering, and Rockefeller) Led organization & moderated ~15 major Tri-I-wide events with industry professionals (100 attendees each) [e.g. "Human Genetics in Drug Development", "Biomedical Innovation in a Non-Profit Organization"] Envisioned, recruited, founded, and led the Symposia Team of 12 new members, in order to develop the first Biotech symposium across the Tri-I (to be held in Fall 2021). Student Leadership, Tri-Institutional CBM PhD Program 2018 - 2021Student Representative, CBM Curriculum Committee (2018); Organizer, Research-in-Progress Seminars (2018-2019); Organizer, CBM Annual Retreat (2019); Head of Social Media (2019 - Present); Summer Internship Admissions Committee (2020) DNA Day Essay Contest - Round 2 Judge, American Society of Human Genetics 2020 2018 - 2019 **Tutor, Grade Expectations** Teach introductory statistics to high school and college students in the NYC area Abstract Committee Member, dVRS Research Symposium 2019, Weill Cornell Medicine 2018 - 2019 Pen Pal, Letters to a Pre-Scientist 2018 - 2019 Exchanged monthly letters with 8th grade students interested in biomedical research Moderator, American Society of Human Genetics Annual Meeting (ASHG), San Diego, CA 2018 Platform Session Title: "Scalable Tools to Enable Collaboration and Reproducible Analyses" Grader, BTRY 4840/6840: "Computational Genetics & Genomics", Cornell University 2017 Research Judge, Cornell Undergraduate Research Board Fall Forum 2017 Athlete & Two-Year Captain, Cornell University Alpine Ski Team 2013 - 2017 Biometry & Statistics Peer Advisor, Cornell Dept Biological Statistics & Computational Biology 2016 - 2017 Cornell Campus Ambassador, Regeneron Pharmaceuticals, Inc. 2016 - 2017 Chair of the New Student Orientation Team, Cornell Hillel 2014 - 2016

Decision: not recommended that ASHG develop a certificate program in suggested topic area(s) at this time.

## **Awards**

Charles J. Epstein Trainee Award for Excellence in Human Genetics Research (Semi-Finalist), American Society of Human Genetics Annual Meeting 2022 (\$990)	2022		
Stanford School of Medicine - Dean's Postdoctoral Fellowship (\$32,784)			
"Reviewer's Choice" award – scoring in the top 10% of all abstract submissions, American Society of Human Genetics Annual Meeting 2020	2020		
NHGRI Scholarship from Keystone Symposia, Beyond a Million Genomes: From Discovery to Precision Health conference (\$1200)	2020		
Charles J. Epstein Trainee Award for Excellence in Human Genetics Research (Semi-Finalist), American Society of Human Genetics Annual Meeting 2019 (\$960)			
"F1000 Outstanding Presentation Prize" for best poster, New York Academy of Sciences: Translating Genetics into Medicine Conference (\$1,000)	2019		
Finalist, Cornell Health Hackathon	2019		
NIH Institutional National Research Service Award (T32), NIGMS - T32GM083937 (\$84,000)	2017-2019		
Genomic Scholars Program, Cornell Center for Vertebrate Genomics (\$5,000)			
Conference Travel Grant, Cornell University - American Society of Human Genetics conference (\$440)			
Conference Travel Grant, Cornell University – Population, Evolutionary, and Quantitative Genetics Conference (\$360)	2018		
Robert S. Elster Memorial Scholarship – for "exhibiting leadership and dedication toward providing outstanding community service to his local community" (\$1,500)	2017		
Dean's List (7x)	2013-2017		
Intern B&B Speaker, Regeneron Pharmaceuticals, Inc. – one of 6 interns nominated to present summer research in a company-wide telecasted presentation	2016		
Special Recognition for Outstanding Geospatial Sciences Exhibit, 4-H Club, New York State Fair.			

# **Completed Upper-Level Graded Coursework (selected)**

Stanford University: Genetics, Ethics, and Society (a 10-week immersive course led by graduate students)

Weill Cornell Medical College: Computational Systems Biology; Clinical and Research Genomics; Genomic Innovation

Cornell University: Computational Genomics; Quantitative Genetics; Human Genomics; Population Genetics; Precision and Genomic Medicine; Bioinformatics Programming; Bayesian Machine Learning; Data Mining and Machine Learning; Probability Models & Inference; Statistical Computing; Linear Models with Matrices; Theory of Statistics; Biological Statistics I & II; Data Structures and Object-Oriented Programming; Physics I & II

# Consortium Membership

dGTEx (Developmental Genotype-Tissue Expression project –		
Assays Tech & Analysis Working Group)		
IGVF (Impact of Genomic Variation on Function – Immune Focus Group, Non-coding FG, Phenotypic Impact FG)	2022 - Present	
ENCODE4 (Variant to Function WG)	2021 - 2023	

## **Peer Review**

Nature Communications
Human Genetics and Genomics Advances
Journal of Crohn's and Colitis
BMC Genomics
Frontiers in Genetics

# **Mentorship**

Years	Name	Type	Institution	Program
2021-23	Pelin (Lin) Poyraz	Undergrad	Cornell University	Biological Sciences, Computational Biology
2022-23	Jon Bezney	PhD rotation	Stanford University	Genetics
2023-	Omar Johnson	Visiting PhD	UTMB/Stanford	MD/PhD Combined Degree Program
2022	Gyu Kim	PhD rotation	Stanford University	Biochemistry
2021-22	Jiayu Liang	Masters	Cornell University	Applied Statistics

# **Professional Organization Membership**

American Society of Human Genetics	2018 - Present
Genetics Society of America	2018 - 2019
New York Academy of Sciences	2017 - 2021

## References

Stephen Montgomery - Stanford University - smontgom@stanford.edu (Postdoc PI)

Andy Clark - Cornell University - ac347@cornell.edu (PhD Co-PI)

Olivier Elemento - Weill Cornell Medicine - ole2001@med.cornell.edu (PhD Co-PI)

Philipp Messer - Cornell University - pm544@cornell.edu (Undergrad PI)

Ana Cvejic - University of Copenhagen - ana.cvejic@bric.ku.dk (Postdoc External Collaborator)

## Other Interests

Lifetime skier (previous captain of Cornell's alpine ski team); Avid runner (1 marathon; 6 half-marathons; NYC Marathon 2022 finisher & 2019 volunteer); Passionate New York sports team fan; Intramural softball; Hiking; Designed a baseball simulation (based on the MLB Showdown card game) as a side project