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EDUCATION

University of Pennsylvania
Cellular and Molecular Biology Doctoral Program: Genetics and Epigenetics

Philadelphia, PA
Fall 2017 Cohort

Northeastern University
Bachelor of Science in Neuroscience, Magna Cum Laude

Boston, MA
Sep 2012- May 2017

AWARDS AND HONORS

- Ruth L. Kirschstein National Research Service Award Pre-doc Fellowship (NRSA F31) 2020-2023
- Northeastern University Class of 2017 Valedictorian
 - <https://www.youtube.com/watch?v=o2C-L3taMIs>
- Northeastern University Dean's List 2013-2017
- Recipient of Greg Jarvis Memorial Scholarship 2017
- Forbes Under30 Summit University Scholar 2016
- Nu Rho Psi National Neuroscience Honors Society 2016-2017
- Tri Beta National Biology Honors Society 2015-2017
- Northeastern University Achievement Award Scholarship 2012-2017

PEER-REVIEWED RESEARCH PUBLICATIONS

- [11] **Singhal P**, Beaulieu-Jones B, Ritchie MD. Leveraging EHR data for disease modeling: a review. **{2023} Manuscript in preparation.**
- [10] **Singhal P**, Kumar R, Morse C, et al. Clinical event prediction in EHR using disease trajectory modeling for cardiovascular outcomes. **{2023} Manuscript in preparation.**
- [8] **Singhal P**, Veturi S, et al. **(2023)** Largest genome-wide long range interaction study identifies interchromosomal associations across 5 complex diseases in the UK Biobank and eMERGE datasets. **Manuscript under review** at American Journal for Human Genetics. Preprint: <https://doi.org/10.1101/2022.10.19.22280888>
- [7] **Singhal P**, Verma SS, Ritchie MD. **(2023)** Gene Interactions in Human Disease Studies – Evidence is Mounting. **Manuscript accepted.** Annual Review of Biomedical Data Science.
- [9] **Singhal P**, Guare L, Morse C, et al. **(2022)** DETECT: Feature extraction method for disease trajectory modeling. **Manuscript accepted.** 16 Dec **2022**. American Medical Informatics Association Informatics Summit. Preprint: <https://doi.org/10.1101/2022.11.06.22281817>
- [6] Levin MG, Tsao NL, **Singhal P**, et al. Multi-ancestry and Multivariate Genome-Wide Analyses Highlight the Role of Common Genetic Variation in Cardiac Structure, Function, and Heart Failure-related Traits. *Nature Communications*. **2022** Sept 22. <https://doi.org/10.1038/s41467-022-34216-6>.
- [5] Gyujoon Hwang, Junhao Wen, Susan Sotardi, Edward S. Brodtkin, Ganesh B. Chand, Dominic B. Dwyer, Guray Erus, Jimit Doshi, **Pankhuri Singhal**, et al. Three Imaging Endophenotypes Characterize Neuroanatomical Heterogeneity of Autism Spectrum Disorder. *JAMA Psychiatry*. **2022** Nov 28. <https://doi.org/10.1101/2022.06.17.22276543>
- [4] Ganesh Chand, **Pankhuri Singhal**, Dominic Dwyer, et al. Schizophrenia imaging signatures and their associations with cognition, psychopathology, and genetics in the general population. *American Journal of Psychiatry*. **2022** Apr 12. <https://doi.org/10.1176/appi.ajp.21070686>

- [3] Nosheen Reza, William P Bone, **Pankhuri Singhal**, et al. (2022) A supervised learning method for classification of electronic health-record based phenotypes. **Manuscript in preparation.**
- [2] Nguyen M, **Singhal P**, Monaghan JR, et al. (2017) Retinoic acid receptor regulation of epimorphic and homeostatic regeneration in the axolotl. *Development*. 144: 601-611. <https://doi.org/10.1242/dev.139873>
- [1] Friedland AE, Baral R, **Singhal P**, et al. (2015) Characterization of Staphylococcus aureus Cas9: a smaller Cas9 for all-in-one adeno-associated virus delivery and paired nickase applications. *Genome Biology* 16: 257. <https://doi.org/10.1186/s13059-015-0817-8>

RESEARCH AWARDS AND GRANTS

- CHARGE Consortium Meeting Travel Award** Awarded July 2022
Awarded by the CHARGE consortium for meeting in Seattle, WA October 12-14, 2022
- Ruth L. Kirschstein National Research Service Award Pre-doc Fellowship (NRSA F31)** Awarded September 2020
Awarded by National Institute for Aging (#F31AG069441-01)
- Genetic and Evolutionary Computation Conference Student Travel Award** Awarded July 2019
Awarded by the special interest group SigEvo of Association for Computing Machinery
- The Provost's Undergraduate Research and Creative Endeavors Award** Awarded Nov 2015
Awarded by the Northeastern University Provost Office
- Research grant awarded in the amount of \$3000 for my proposal: "*CRISPR/Cas9 mediated knockout of novel genes regulating limb patterning in the regenerating axolotl model.*" The project was conducted in the lab of Dr. James Monaghan as a directed study in 2015-2016.
- The Provost's Undergraduate Research and Creative Endeavors Award** Awarded Nov 2014
Awarded by the Northeastern University Provost Office
- Research grant awarded in the amount of \$2000 for my proposal: "*Targeting the Retinoic Acid cell signaling pathway during regeneration in the axolotl model to elucidate limb patterning.*" The project was conducted in the lab of James Monaghan as a directed study in 2014-2015.

GRADUATE RESEARCH EXPERIENCE

- University of Pennsylvania, Department of Genetics** Philadelphia, PA
Genetics and Epigenetics Doctoral Student May 2018 - Present
Dissertation Research: Lab of **Dr. Marylyn D. Ritchie**, Professor, Director of Center for Translational Bioinformatics
- **Thesis work:**
 - *Disease Trajectory Project:* Developing longitudinal data analysis methods to integrate patient electronic health records for clinical event prediction using Penn EHR data
 - *Epistasis in disease architecture:* Tested association between 5.2 million long-range/high LD epistatic models with complex diseases using UKBB and eMERGE data
 - **Collaborations:**
 - Lead a collaboration with Penn Neuroimaging group to identify genetic basis for depression, ASD, schizophrenia. Conducted GWAS and PRS
 - Collaborated with Penn Cardiology to implement unsupervised clustering algorithms in heart failure population to identify HF subtypes using phenomic data
 - **Talks:**
 - CHARGE consortium webinar, Feb meeting; Feb 8, 2023 (virtual)
 - Mt. Sinai Institute for Genomic Health – EHR Working Group; Jan 19, 2023 (NY)
 - CHARGE Consortium Meeting; Oct 14, 2022 (Seattle) – Blitz Talk
 - Pacific Symposium on Biocomputing; January 3, 2023 (Big Island, Hawaii) – "*Tracing patient disease trajectories in electronic health records*" in workshop "High-Performance Computing Meets High-Performance Medicine"
 - *Upcoming:* AMIA Informatics Summit; March 13-16, 2023 (Seattle) – "*Disease trajectory modeling in Penn Medicine EHR*"
 - **Other:**

- Co-chair of session SALUD: Scalable Applications of cLinical risk Utility and preDiction for Pacific Symposium on Biocomputing conference January 3-6, 2023 (Big Island, Hawaii)
- Reviewer for journal *BioData Mining*

Select poster sessions:

- [9] Pankhuri Singhal, Lindsay Guare, Anastasia Lucas, Colleen Morse, Marta Byrska-Bishop, Marie A. Guerraty, Dokyoon Kim, Marylyn D. Ritchie, and Anurag Verma. Defining longitudinal disease trajectories in 146,000 individuals with hypertension from Penn Medicine EHR. **ASHG**; 2022 October 25-29 (LA) ***author's choice abstract award**
- [8] **Pankhuri Singhal**, Lindsay Guare, Anastasia Lucas, Colleen Morse, Marta Byrska-Bishop, Marie A. Guerraty, Dokyoon Kim, Marylyn D. Ritchie, and Anurag Verma. Identifying longitudinal disease trajectories and their clinical associations in 146,000 individuals with hypertension from Penn Medicine Electronic Health Records. **CHARGE Meeting** ; 2022 October 12-14 (Seattle)
- [7] **Pankhuri Singhal**, Anurag Verma, Dokyoon Kim, Marylyn D. Ritchie. Clinical event prediction in complex genetic traits leveraging longitudinal EHR and genomic data in Penn Medicine BioBank. **Pacific Symposium on Biocomputing**; 2022 January 3-6 (Big Island)
- [6] **Pankhuri Singhal**, Shefali S. Verma, Anastasia Lucas, Sudha Veturi, Alex T. Frase, Steven Schrodi, David Fasel, Chunhua Weng, Sarah Pendergrass, Daniel J. Schaid, Iftikhar J. Kullo, Ozan Dikilitas, Patrick M.A. Sleiman, Hakon Hakonarson, Marylyn D. Ritchie. Genome-wide inter-chromosomal epistatic associations identified across complex diseases in the ~300,000 participants from eMERGE and UK Biobank. **ASHG**; 2021 October 18-22 (Virtual) ***author's choice abstract award**
- [5] **Pankhuri Singhal**, Marie A. Guerraty, Dokyoon Kim, Daniel J. Rader, Marylyn D. Ritchie, and Anurag Verma. Determining disease co-occurrence architecture of Hypertensive Heart Disease in Penn Medicine Biobank using longitudinal EHR data linked with PMBB participants. **American Heart Association**; 2021 November 13-15 (Virtual)
- [4] **P. Singhal**, S.S. Verma, M.D. Ritchie. Leveraging evolutionary drivers of genetic variation reveals complexity underlying diseases across eMERGE and UKBioBank. **MidAtlantic Bioinformatics Conference**; 2020 November 2 (Virtual)
- [3] **P. Singhal**, J.E. Miller, A. Verma, S.S. Verma, S.M. Dudek, M.D. Ritchie. Neural Network-based multiomics data integration in Alzheimer's Disease. **ASHG**; 2019 October 14-19 (Houston, TX)
- [2] **P. Singhal**, J.E. Miller, A. Verma, S.S. Verma, S.M. Dudek, M.D. Ritchie. Neural Network-based multiomics data integration in Alzheimer's Disease. Poster session at **Genetic and Evolutionary Computation Conference**; 2019 July 13-17 (Prague, Czech Republic)
- [1] **P. Singhal**, S.S. Verma, J.E. Miller, S.M. Dudek, M.D. Ritchie. Using simulation studies to evaluate the effect genetic architecture has on neural network-based predictions. Poster session at **MidAtlantic Bioinformatics Conference**; 2018 October 29 (Philadelphia, PA)

TEACHING /MENTORING EXPERIENCE

G&E Peer Mentorship program

2019 - Present

- I am a peer mentor as part of the CAMB G&E program, I meet with incoming students assigned to me on a need basis to help them navigate coursework, selecting rotations, deciding on a thesis lab, and working through general grad school/Philly challenges.

Shine in Math Academy - SAT Prep + Essay Writing Teacher

May 2017 - Present

- Tutor highschoolers in essay-writing and SAT prep over zoom sessions monthly for college admissions. Created various workshops to help high school students develop strong writing habits and develop their own literary style and voice. Conduct 1:1 ideation sessions to help students think creatively about writing topics. Tutor in summer 8 week Math SAT prep course annually.

GWAS + PRS (Penn CBICA seminar) Workshop

August 2021

- I was invited to lead an interactive 4 hour workshop on running GWAS and PRS for the CBICA's seminar series entitled "Endophenotyping III: Genetics research in Action". I worked with machine learning scientists and neuroimaging researchers to teach them how to leverage imaging data to conduct genome wide association studies and polygenic risk scores.

Summer Undergraduate Internship Program Mentor (SUIP)

Summer 2021

- I was a mentor for an SUIP scholar in our lab teaching her bioinformatics and computational skills to run genomic analyses. Specifically, Alexis's 10-week project focused on learning unix and R to maneuver around our computing cluster and conduct data pre-processing (weeks 1-5), running data QC for UKBB genotype array data (weeks 6-7), running a GWAS (week 8), running a PRS (week 9), and writing up a poster (week 10) for the SUIP summer conference.

Penn Genetics Summer Internship Program Mentor (PGSIP)

Summer 2021

- Co-mentor for summer undergraduate student Anna in the genetics department; weekly meetings
- Helped pilot mentorship program for incoming PhD students. Volunteered at three-day workshop to help prepare students for graduate school challenges in the lab, academic, and otherwise

American Physician Scientist Association Summer Program Mentor (APSA)

Summer 2020

- I was a mentor for and APSA scholar in our lab, teaching him how to conduct genomic analyses (GWAS, PRS) in large datasets such as UKBB to understand whether socioeconomic variables played a role in cardiovascular events. Specifically, in the 2 month program, David learned how to do data pre-processing and QC for GWAS and analyzed his results using pathway analysis tools. He presented his work at the APSA poster session.

PRE-GRADUATE SCHOOL RESEARCH EXPERIENCE

Northeastern University Regenerative Biology Laboratory – Monaghan Lab

Boston, MA

Undergraduate Researcher

Jan 2014- May 2017

Advisor: Dr. James Monaghan, Assistant Professor

- Investigated cellular and molecular underpinnings of tissue regeneration in the axolotl salamander. Elucidated the transcriptional proximal-distal (PD) limb patterning duplication that occurs during regeneration when targeting the retinoic acid (RA) cell signaling pathway using microarray analysis in time-course experiment. Validated genes via CRISPR/Cas9 gene-editing system. Skills: cloning, in-situ hybridization, IHC, whole mount staining, and histology, invasive surgery, axolotl husbandry.

○ **Publications:**

Nguyen M, **Singhal P**, Monaghan JR, *et al.* (2017) Retinoic acid receptor regulation of epimorphic and homeostatic regeneration in the axolotl. *Development*. **144**: 601-611

○ **Posters:**

- 1) M. Nguyen, **P. Singhal**, S.R. Voss, M. Maden, J.R. Monaghan. Molecular encoding of positional information during limb regeneration. Poster session at: 3rd Annual *Research, Innovation and Scholarship Expo (RISE)*; 2015 April 9; Northeastern University, Boston, MA.
- 2) M. Nguyen, **P. Singhal**, S.R. Voss, M. Maden, J.R. Monaghan. Molecular encoding of positional information during limb regeneration. Poster session at: 75th Annual *Society for Developmental Biology (SDB) Meeting* 2016 August 4-7; Boston, MA.

Editas Medicine

Cambridge, MA

Molecular Biology Co-op, Platform and Disease-Indication

(full-time employee) Jan 2016-Aug 2016

Advisor: Dr. Ari Friedland, Scientist

- Cloned CRISPR/Cas9 gRNA constructs targeting Herpes Simplex Virus type 1 (ocular keratitis) for in vitro studies via transfection in human cell lines. Engineered adeno-associated virus (AAV2) vectors for in-vivo mouse studies to measure viral reactivation upon editing. Optimized vector configurations by testing tissue-specific promoters and gRNAs. Determined cutting efficiency via molecular mutation assay.
- Developed inactivating anti-Cas system to regulate catalytic activity of Cas9, in-vitro via AAV delivery. Targeted CCR5 locus, optimized vector configurations via FACS readout.

○ **Posters:**

- 1) **P. Singhal**, A. Sadowski, R. Baral, A. Friedland, D. Bumcrot. Inactivating anti-cas in gene editing. Poster session at: 20th Annual *American Society of Gene and Cell Therapy (ASGCT) Meeting*; 2017 May 10-13; New Orleans, LA.
- 2) C. Owens, B. Diner, R. Fusco, E. King, A. Friedland, **P. Singhal**, K. Gogi, F. Harbinski, S. Shen, M. Stefanidakis, L. Barrera, D. Bumcrot, D. Neumann, C. Albright. CRISPR/Cas9 Targeted Disruption of Herpes Simplex Virus type 1 in a Rabbit Latency Model Reduces Viral Reactivation and Associated Corneal Pathology. Poster session at:

9th Annual Association for Research in Vision and Ophthalmology (ARVO) Meeting; 2018 April 28-May3;
Honolulu, HI.

Editas Medicine

Molecular Biology Co-op, Platform and Disease-Indication
Advisor: Dr. Ari Friedland, Scientist

Cambridge, MA

(full-time employee) Jan 2015-Aug 2015

- Characterized *Staphylococcus aureus* Cas9 as an effective nuclease alternative for gene-editing in the CRISPR/Cas9 gene-editing system. Given smaller viral packaging size, showed that an all-in-one adeno-associated virus (AAV) multiplexing approach with *S. aureus* using dual nickases D10A and N580A could promote homology directed repair (HDR) at endogenous loci. Assessed cleavage efficacy of gRNAs by T7 Endonuclease 1 molecular mutation assay via transfection in human cell line. Conducted TOPO-cloning of the target loci in order to characterize indel profile due to HDR. Evaluated off-targets by performing and optimizing GUIDE-Seq assay. Used luciferase reporter assay to quantify functional knock-down of genes to compare gRNA cleavage efficiency. Assessed results via flow-cytometry.

○ Publications:

A.E. Friedland, S. Shen, **P. Singhal**, A. Sousa, M. Collins, M.L. Maeder, G.G. Welstead, H. Jayaram, D. Bumcrot. *Staphylococcus aureus* Cas9 an alternative Cas9 for genome engineering applications. Poster session at: 18th Annual American Society of Gene and Cell Therapy (ASGCT) Meeting; 2015 May 13-16; Washington, D.C.

EDITORIAL WORK

Freelance Editor/Founder “JusttheWriteWord”

Jan 2013-Present

- I work as a literary/editorial mentor through an organization I founded called JusttheWriteWord in 2013 (<https://pankhurisinghal.com/editorial>). I work primarily with high school students applying to college and post-grad students applying for higher education. I have worked with students in person and remotely to do any of the following: ideate their narrative, refine/edit existing work, develop creative writing techniques, identify literary voice through writing exercises, etc.
- Content and copy edit manuscripts pre-publication. Cited as Primary Editor of the following three books:
 - 1) Rao, Rupen. *Ayurveda Cookbook: Eating for Mind, Body, and Consciousness*. Washington, D.C.: Warren Publishing, 2015. 1st Edition. Print.
 - 2) Rao, Rupen. *Indian Cooking: Popular Restaurant Dishes*. Washington, D.C.: Warren Publishing, 2014. 1st Edition. Print.
 - 1) Rao, Rupen. *Indian Cooking: From My Mom*. Washington, D.C.: Warren Publishing, 2013. 1st Edition. Print.
 - 2) Chiaravolleti, Isabella J. *Milk for No One*. Createspace Independent Publishing Platform, 2017. 1st Edition. Print.

PERSONAL

- Vocalist – Berklee College of Music Indian Ensemble (2014-2017)
 - 2023 **Grammy Nominee** for Best Global Album “Shuruaat” (ensemble vocalist)
- Scuba diving, poetry, hiking, surfing, oil pastel painting