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**CONTACT**

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**EDUCATION**

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

Philadelphia, PA  
Aug 2017- April 2023

**Northeastern University**  
Bachelor of Science in Neuroscience, Magna Cum Laude

Boston, MA  
Sep 2012- May 2017

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**AWARDS AND HONORS**

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- 2023 **Grammy**® Nominated vocalist on album 'Shuruaat' in Best Global Album category
  - <https://ffm.to/shuruaat>
- Northeastern University Class of 2017 **Commencement Speaker**
  - [https://youtu.be/o2C-L3taMIs?si=X0By\\_9RUQdkVfviP](https://youtu.be/o2C-L3taMIs?si=X0By_9RUQdkVfviP)
- 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

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**RESEARCH AWARDS AND GRANTS**

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**NRSA T-32 Postdoctoral Fellowship** Awarded July 2023  
Awarded NRSA T-32 Genomic Medicine Fellowship (University of Pennsylvania)

**SAIL Meeting Travel Award** Awarded Feb 2023  
Awarded by the Symposium on AI for Learning Health Systems program committee

**CHARGE Consortium Meeting Travel Award** Awarded July 2022  
Awarded by the Cohorts for Heart and Aging Research in Genomic Epidemiology consortium

**Ruth L. Kirschstein National Research Service Award Pre-doc Fellowship (NRSA F31)** Awarded Sept 2020  
Awarded by National Institute for Aging #F31AG069441-01 (3-year duration)

**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.

## PEER-REVIEWED RESEARCH PUBLICATIONS

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- [15] **Singhal P**, Kumar R, Kumar N, Becker M, Hanish A, Xapakhdly K, Verma A. Leveraging GPT models to intuitively structure free-text clinical notes. **(2023)**. Accepted to *American Medical Informatics Summit*.
- [14] Lee DSM, DePaolo JS, Aragam KG, Biddinger K, Conery M, Dilitikas O, Hoffman-Andrews L, Judy RL, Khan A, Kulo I, Puckelwartz MJ, Reza N, Satterfield BA, **Singhal P**, et al. Common- and rare-variant genetic architecture of heart failure across the allele frequency spectrum. **(2023)**. *Nature Medicine*. 10.1101/2023.07.16.23292724
- [13] **Singhal P**, Tan A, Drivas T, Ritchie MD, Beaulieu-Jones B. Opportunities and Challenges for Biomarker Discovery Using Electronic Health Record Data. **(2023)** 18 July 2023. *Trends in Molecular Medicine*.  
<https://doi.org/10.1016/j.molmed.2023.06.006>
- [12] **Singhal P**, Verma SS, Ritchie MD. **(2023)** Gene Interactions in Human Disease Studies – Evidence is Mounting. 17 May 2023. *Annual Review of Biomedical Data Science*. <https://www.annualreviews.org/doi/10.1146/annurev-biodatasci-102022-120818>
- [11] **Singhal P**, Guare L, Morse C, et al. **(2023)** DETECT: Feature extraction method for disease trajectory modeling. 16 June 2023. *American Medical Informatics Association Informatics Summit*.  
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC10283148/>
- [10] **Singhal P**, Veturi S, Dudek SM, et al. **(2023)** Evidence of epistasis in regions of long-range linkage disequilibrium across five complex diseases in the UK Biobank and eMERGE datasets. 6 Apr 2023. *American Journal for Human Genetics*. <https://doi.org/10.1016/j.ajhg.2023.03.007>
- [9] **Singhal P**, Veturi Y, Judy R, Park Y, Vujkovic M, Veatch O, Kember R, Verma SS. **(2023)** session Introduction: SALUD: Scalable Applications of cLinical risk Utility and preDiction. *Pacific Sym on Biocomputing*.  
[https://doi.org/10.1142/9789811270611\\_0037](https://doi.org/10.1142/9789811270611_0037)
- [8] Nam Y, Jung S, Yun J, Sriram V, **Singhal P**, et al. **(2023)** Discovering comorbid diseases using an inter-disease interactivity network based on biobank-scale PheWAS data. *Bioinformatics*.  
<https://doi.org/10.1093/bioinformatics/btac822>
- [7] Hwang G, Wen J, Sotardi S, Brodtkin ES, Chand GB, Dwyer DB, Erus G, Doshi J, **Singhal P**, et al. **(2023)** Assessment of Neuroanatomical Endophenotypes of Autism Spectrum Disorder and Association With Characteristics of Individuals With Schizophrenia and the General Population. *JAMA Psychiatry*. doi:10.1001/jamapsychiatry.2023.0409
- [6] Kumar R, **Singhal P**, Guare L, Morse C, Byrska-Bishop M, Ritchie MD, Verma A. **{2023}** Predicting cardiovascular outcomes and disease trajectories using recurrent neural networks and probabilistic graphs. **Manuscript in preparation.**
- [5] Levin MG, Tsao NL, **Singhal P**, et al. **(2022)** Genome-wide association and multi-train analyses characterize the common genetic architecture of heart failure. *Nature Communications*. <https://doi.org/10.1038/s41467-022-34216-6>
- [4] Chand G, **Singhal P**, Dwyer D, et al. **(2022)** Schizophrenia imaging signatures and their associations with cognition, psychopathology, and genetics in the general population. *American Journal of Psychiatry*.  
<https://doi.org/10.1176/appi.ajp.21070686>
- [3] Reza N, Bone WP, Yang Y, **Singhal P**, et al. **{2022}** A supervised learning method for classification of electronic health-record based phenotypes. Preprint. <https://doi.org/10.1101/2022.10.31.22281772>
- [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>

## GRADUATE/POSTDOC RESEARCH EXPERIENCE

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**University of Pennsylvania, Department of Medicine**  
T32 Genomic Medicine Postdoctoral Fellow

Philadelphia, PA  
June 2023 – Present

- I am completing a short-term postdoc in the lab of **Dr. Anurag Verma** focused on leveraging LLMs for identification of undiagnosed patients with rare diseases like Marfan's syndrome in the Penn Medicine BioBank in collaboration with Argonne National Labs

- **Professional opportunities:**
  - Panelist for “LLMs in EHR” session at Institute for Biomed Informatics retreat; Bear Creek, PA (Sept. 5, 2023)
  - Serve as Proceedings subchair for Health, Inference, and Learning (CHIL) 2024
  - Lead an inter-departmental large language model working group across the Institute of Biomedical Informatics (Penn) with biweekly presentations, tutorials, and workshops; presented at Ritchie Symposium (Jan 2024, Phil.)
  - I lead and organize a *Digital Health Working Group* (~30 individuals) at the 2023 NeurIPS conference; we are currently working on a review paper together
  - Serve as reviewer for journals: *Genome Biology*, *BioData Mining*, *Amer. Journal of Human Genetics*, and *Bioinformatics*
- **Start-up work:**
  - Completed start-up bootcamp with Penn’s NSF funded Penn I-Corp chapter; team co-founder for medical start-up “PredixHealth” (Sept. - Nov 2023)
- **Coursework (auditing):**
  - ESE 3060 Applied Deep Learning (2023 Fall)
  - CIS 5200 Applied Machine Learning (2024 Spring)

**University of Pennsylvania, Department of Genetics**

*Genetics and Epigenetics Doctoral Student*

Philadelphia, PA

Aug 2017 – April 2023

**Thesis Advisor: Dr. Marylyn D. Ritchie**, Professor, Director of Center for Translational Bioinformatics

**Thesis:** “Epistasis and Evolution of Disease Trajectories in Multi-Dimensional Study of Genomic and Phenomic Interactions” ([dissertation PDF](#))

- **Thesis work:**
  - *Modeling disease trajectories in electronic health records:* Developed longitudinal data analysis methods to integrate patient electronic health records for clinical event prediction using Penn EHR data
  - *Detecting epistasis in complex disease architecture:* Tested association between 5.2 million long-range/high LD SNP-SNP epistasis models with complex diseases using biobank data
- **Collaborations:**
  - Led a collaboration with Penn Neuroimaging group to identify genetic basis for depression, ASD, schizophrenia; conducted genome-wide association studies and polygenic risk score analysis
  - Collaborated with Penn Cardiology to implement unsupervised clustering algorithms in heart failure (HF) population to identify HF subtypes using phenomic data
- **Talks:**
  - American Medical Informatics Association Informatics Summit, March 14, 2023 (Seattle) – “*Leveraging longitudinal data for disease trajectory modeling using DETECT*”
  - CHARGE consortium webinar, 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”
- **Professional opportunities:**
  - Pacific Symposium on Biocomputing conference: Co-chair of session SALUD: Scalable Applications of cLinical risk Utility and prediction; Jan. 3-6, 2023 (Big Island, Hawaii)

**Select poster sessions:**

[10] **Singhal P**, Kumar R, Kumar N, Becker M, Hanish A, Xapakhdly K, Verma A. A large language model framework to identify patients with undiagnosed rare genetic diseases using clinical notes. **{2024}**. Submitted to ***Symposium on Artificial Intelligence in Learning Health Systems***.

[9] **Pankhuri Singhal**, Lindsay Guare, Rachit Kumar, Colleen Morse, Anastasia Lucas, Marta Byrska-Bishop, Marie A. Guerraty, Dokyoon Kim, Anurag Verma, Marylyn D. Ritchie. Disease trajectory modeling and clinical event prediction of hypertension outcomes in Penn Medicine EHR. **Symposium on Artificial Intelligence in Learning Health Systems**; 2023 May 9-12, 2023 (Puerto Rico)

- [8] **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**
- [7] **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)
- [6] **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 (Hawaii)
- [5] **Pankhuri Singhal**, Shefali S. Verma, Anastasia Lucas ... 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**
- [4] **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
- [3] **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)
- [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 Bioinf. Conference**; 2018 October 29 (Philadelphia, PA)

## TEACHING /MENTORING EXPERIENCE

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### T32 Postdoctoral Fellow

Summer 2023

- I am the primary mentor for summer intern Luke Mutz. He is pursuing a masters in statistics at Villanova University and I am currently introducing him to genomic analyses (GWAS, PRS, etc.).

### 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. Currently mentoring Mariah and Aude.

### Shine in Math Academy - SAT Prep + Essay Writing Teacher

May 2017 - Present

- I tutor high school students in essay-writing and SAT prep over zoom sessions monthly for college admissions. I have created various workshops to help 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 David, an 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.

## UNDERGRADUATE RESEARCH EXPERIENCE

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### 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: 3<sup>rd</sup> Annual *Research, Innovation and Scholarship Expo*; 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: 75<sup>th</sup> Annual *Society for Developmental Biology*; 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 and optimized adeno-associated virus (AAV2) vectors for in-vivo mouse studies to measure viral reactivation upon editing.
- 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: 20<sup>th</sup> 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: 9<sup>th</sup> Annual *Association for Research in Vision and Ophthalmology (ARVO)* Meeting; 2018 April 28-May3; Honolulu, HI.

### Editas Medicine

Cambridge, MA

*Molecular Biology Co-op, Platform and Disease-Indication*

*(full-time employee)* Jan 2015-Aug 2015

Advisor: Dr. Ari Friedland, Scientist

- 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.

#### 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: 18<sup>th</sup> Annual  
American Society of Gene and Cell Therapy (ASGCT) Meeting; 2015 May 13-16; Washington, D.C.

## EDITORIAL WORK

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### Freelance Editor/Founder *JusttheWriteWord*

Jan 2013-Present

- I work as a freelance literary editor to content and copy edit manuscripts pre-publication. I am passionate about mentoring students in writing. I founded an organization called JusttheWriteWord in 2013 (<https://pankhurisinghal.com/editorial>), through which I work with writers of all backgrounds. I have worked with students applying to college and post-grad programs (med school, MBA, masters, PhD programs), as well as established authors. I tailor the curriculum to the writer and the stage they are in for any or all of the following: ideating narrative, refining existing work, developing creative writing techniques, identifying literary voice through writing exercises, etc.
- I am the editor of the following books:
  1. Chiaravolleti, Isabella J. *Milk for No One*. Createspace Independent Publishing Platform, 2017. 1<sup>st</sup> Edition. Print.
  2. Rao, Rupen. *Ayurveda Cookbook: Eating for Mind, Body, and Consciousness*. Washington, D.C.: Warren Publishing, 2015. 1<sup>st</sup> Edition. Print.
  3. Rao, Rupen. *Indian Cooking: Popular Restaurant Dishes*. Washington, D.C.: Warren Publishing, 2014. 1<sup>st</sup> Edition. Print.
  4. Rao, Rupen. *Indian Cooking: From My Mom*. Washington, D.C.: Warren Publishing, 2013. 1<sup>st</sup> Edition. Print.

## PERSONAL

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- **Philadelphia Improv Theater** (2023 – Present)
  - **Improv Performer** on team *Pass the Kiss* (founding member); perform at venues around Philadelphia
  - Completed Levels 1-3 of training at Philadelphia Improv Theater (PHIT)
- **Kelly Writers House** (2022-Present)
  - **Poet**; meet monthly as part of the oldest poetry club at UPenn to share poetry and perform poetry + spoken word at local readings
- **Berklee College of Music Indian Ensemble** (2014-2017)
  - **Ensemble Vocalist**; classically trained in Hindustani vocal music
  - Honored to have been a 2023 **Grammy® Nominee** for Best Global Album “Shuruaat”; assembled our 10-year commemorative album with my talented ensemble-mates (<https://open.spotify.com/album/54EDAsIzpTmJ0e8tm4We92>)
- **Passions**: scuba diving, poetry, songwriting, singing, improv, ultimate frisbee, salsa dancing, hiking, surfing, abstract seascape painting, fossil hunting