

SAYAN GHOSAL

Cambridge * ghosal@broadinstitute.org * <https://sayangsep.github.io> * LinkedIn * 443-531-5268

PROFESSIONAL SUMMARY

- Computational scientist with a passion for integrating complex statistical models with genomic knowledge to provide insight into disease mechanisms.
- Contributions include novel Bayesian and machine learning models providing mechanistic insights into complex disorders like Alzheimer's, Autism, and Schizophrenia.
- Highly collaborative and motivated to drive new research endeavors in the intersection of methods development and biological discovery.

ACADEMIC BACKGROUND

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| Johns Hopkins University , Baltimore, USA | |
| Ph.D. , Electrical and Computer Engineering | 2023 |
| M.S. , Applied Mathematics and Statistics | 2021 |
| Jadavpur University , Kolkata, INDIA | |
| B.E. , Electronics and Telecommunication Engineering | 2017 |

PROFESSIONAL BACKGROUND

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| Computational Scientist, Broad Institute of MIT and Harvard , Cambridge | <i>Present</i> |
| <i>Contributions: Graph-Based SV Discovery, Long/Short Read Sequencing, Disease Association</i> | |
| AI Resident, Google X , Mountain View | <i>2022</i> |
| <i>Contributions: Genetics, LLM, Mixed Effect Modelling, Time Series Analysis, HPC</i> | |
| ML Intern, Siemens Healthineers , Princeton | <i>2021</i> |
| <i>Contributions: Graph Neural Networks, Contrastive Learning, Interpretability</i> | |

SKILLS

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| GL for Genomics | Graph neural networks for structural variant calling, graph-attention models for polygenic risk scoring. |
| ML for Genomics | Deep Bayesian models for finemapping, latent-factor models for multimodal imaging-genetics |
| DL for Genomics | Interpretable autoencoder for multi-omics, generative models for comparative transcriptomic, transformers integrating time-series and genetics |
| Statistical Genetics | GWAS, structural variant discovery, PRS analysis, finemapping, imaging-genetics |
| Deep Learning | Contrastive Learning on Graphs, Transformers, Autoencoders, GNN |
| Model Interpretability | Motif Discovery, Bayesian Feature Selection, Attention Mechanisms, LASSO, Group-LASSO |

RELEVANT EXPERIENCE

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| Broad Institute , Cambridge | <i>Present</i> |
| <i>Computational Scientist</i> | |
| Deep generative models for comparative transcriptomics | |
| • Developing diffusion models for cross-species single cell genomics data. | |

- Transfer learning the effects of disease or drugs in single-cell expressions from animal models to humans.

Motif Driven Structural Variant Discovery

- Identifying structural variants from the graph representation of sequence alignments.
- Finding novel motifs for complex genomic rearrangements.

Johns Hopkins University, Baltimore

2017-2023

Research Assistant, Electrical and Computer Engineering

BEATRICE: Bayesian Fine-mapping from Summary Data using Deep Variational Inference

- Developed a deep Bayes variational approach to parse complex heritability resulting in 2.2 fold increase in power and coverage.
- Utilized machine learning with Bayesian inference to handle multiple causal variants and infinitesimal effects from non-causal variants.

A Biologically Interpretable and Non-linear Approach to Generate Polygenic Risk Scores

- Consolidated genetic risk along biological pathways to generate risk scores predictive of disorder.
- Embedded gene ontology in a graph to infer underlying processes and functions linked to disease risk prediction.

A Biologically Interpretable Graph Convolutional Network to Link Genetic Risk Pathways and Neuroimaging Markers of Disease

- Developed a novel geometric deep learning tool for whole-brain whole-genome analysis of schizophrenia.
- Collaborated with cross-functional teams of biologists, data scientists, and clinicians, which led to a future million-dollar grant, scholarships², awards¹, and two publications.

Multimodal Imaging Genetics Models for Biomarker Identification and Schizophrenia Risk Prediction

- Developed novel latent factor models utilizing autoencoder and dictionary learning to identify correlated brain and genetic networks from brain imaging and genetics study of schizophrenia.
- Received special mention in the Hopkins magazine and a best paper award³ at SPIE.

SUPERVISING ACTIVITY

Johns Hopkins University, Baltimore

2021- 2023

Supervisor

- Advising a computer science graduate student on deep learning projects aimed to learn the longitudinal effect of genetic variations on morphological changes in brain regions of 1K Alzheimer's patients.
- Authored a senior author paper at the International Conference of the IEEE Engineering in Medicine and Biology Society.

HONORS AND AWARDS

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| ¹ Organization for Human Brain Mapping awarded \$700 for noteworthy abstracts. | 2023 |
| ² MINDS fellowship awarded \$30K for spring tuition. | 2022 |
| ³ Best Paper Award, SPIE Medical Imaging (Image Processing Conference) | 2021 |
| ⁴ MICCAI travel award of \$500. | 2020 |
| ⁵ Dept. of Electrical and Computer Engineering, JHU, PhD fellowship | 2017-2018 |
| ⁶ Mitacs Globalink Research Fellowship Award | 2016 |

PATENTS

Ghosal, S., Jacob, A. J., Sharma, P., & Gulsun, M. A. (2023). Subpopulation Based Patient Risk Prediction Using Graph Attention Networks. US Patent App. 17/647,613.

PUBLICATIONS

S. Ghosal, et al., *GUIDE-PRS: A Biologically Interpretable and Non-linear Approach to Generate Polygenic Risk Scores.*(In Prep).

S. Ghosal, et al., *BEATRICE: Bayesian Fine-mapping from Summary Data using Deep Variational Inference.*(Submitted in *PLOS Genetics*). biorXiv

S. Wu, A. Venkataraman, S. Ghosal. *GIRUS-net: A Multimodal Deep Learning Model Identifying Imaging and Genetic Biomarkers Linked to Alzheimer's Disease Severity.* EMBC, 2023.

S. Ghosal, et al. *A Biologically Interpretable Graph Convolutional Network to Link Genetic Risk Pathways and Neuroimaging Markers of Disease.* **ICLR: International Conference on Learning Representations, 2022 (Accepted).** biorXiv

S. Ghosal, et al. *A Generative Discriminative Framework that Integrates Imaging, Genetic, and Diagnosis into Coupled Low Dimensional Space.* NeuroImage: 238:118200, 2021

S. Ghosal, et al. *G-MIND: An End-to-End Multimodal Imaging-Genetics Framework for Biomarker Identification and Disease Classification.* Proc. SPIE, Medical Imaging 2021: Image Processing. **Selected for Special Oral Presentation (<15% of Papers), and received best student paper award**

S. Ghosal, et al. *Bridging Imaging, Genetics, and Diagnosis in a Coupled Low-dimensional Framework.*MICCAI: Medical Image Computing and Computer Assisted Intervention, 2019. **Selected for Early Acceptance (Top 18% of Submissions)**

S. Ghosal, et al. *A generative-predictive framework to capture altered brain activity in fMRI and its association with genetic risk: application to Schizophrenia.* Proc. **SPIE** 10949, Medical Imaging 2019: Image Processing.

S. Ghosal, Nilanjan Ray. *Deep deformable registration: Enhancing accuracy by fully convolutional neural net.* **Pattern Recognition Letters.**

S. Ghosal, et al. *A novel non-rigid registration algorithm for zebrafish larval images.* 39th Annual International Conference of the IEEE Engineering in Medicine and Biology Society (**EMBC**), 2017.

INVITED SEMINARS AND TALKS

Title: Benefits of Deep Learning to Parse Complex Genetic Architectures to Provide Mechanistic Insights

MIT (Host: Manolis Kellis)

2023

Title: Deep Imaging Genetics to Parse Neuropsychiatric Disorders

Regeneron (Host: Yu Bai)

2023

Google-Genomics, Google Health (Host: Farhad Hormozdiari)

2022

Title: Biologically Inspired Regularization Models Integrating Multimodal Data to Parse Neuropsychiatric Disorders.

ECE Seminar Series (Host: Archana Venkataraman)

2022