

# SHLOKA NEGI

Ph.D. candidate, University of California, Santa Cruz, USA

Biomolecular Engineering & Bioinformatics

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## EDUCATION

University of California, Santa Cruz (UCSC)

(Sept 2022 – ongoing)

Ph.D. candidate in Biomolecular Engineering and Bioinformatics

Indian Institute of Technology (B.H.U.), Varanasi

(July 2018 – May 2022)

B.Tech (Hons.) in Pharmaceutical Engineering and Technology

CGPA : 9.91/10

## SKILLS AND INTERESTS

**Skills:** Python, R, Bash, WDL, AWK, Snakemake, Docker, Slurm, Toil, Git

**Research Interests:** Computational Genomics, Long-Read Sequencing, Precision Medicine, Pangenomics

## RESEARCH EXPERIENCE

### • PhD Thesis at University of California, Santa Cruz

(Apr'23 - ongoing)

*Proposal*

**Supervisors:** [Dr. Benedict Paten](#) and [Dr. Karen Miga](#), UCSC Genomics Institute

**Collaborators:** Funded by CZI and NIH. Collaboration with The Broad Institute of MIT and Harvard, Children's National Hospital, D.C., and Google Health

**Title:** Advancing diagnostics with scalable long-read nanopore sequencing, assembly, variant calling, and methylation profiling

**Aim 1:** Create a scalable and cost-efficient pipeline using long-read sequencing to identify rare pathogenic variants in undiagnosed rare disease patients. Optimize a one-flow cell sequencing protocol to reduce costs to ~ \$1000 per sample and computational analysis costs to ~ \$100 on a cloud-based platform. Develop automated pipelines for variant-calling, clinical annotation, and identifying potential candidates for de novo calling, compound-heterozygous calling, structural variant annotation, and complex tandem-repeat annotation.

**Aim 2:** Develop methods for methylation analysis of thousands of post-mortem frontal cortex brain tissue samples sequenced by The NIH's Center for Alzheimer's and Related Dementias (NIH CARD). Perform methylation quantitative trait locus (m-QTL) analyses to identify SVs that impact methylation and expression.

**Aim 3:** Develop pangenome-guided genome assembler using long-read mapping to the pangenome. Generate haplotype-resolved assembly of medically relevant complex regions harboring high-identity segmental duplications, such as CYP21A2/CYP11A1P, MHC, SMN1/2 genes, and many more involved in rare diseases.

### • Research Internship at Fraunhofer, Germany (DAAD-WISE Scholarship)

(Aug'21-Sep'21)

*Project, Report*

**Supervisor:** [Dr. Martin Hoffmann](#), Experimental Medicine and Therapy Research, Universität Regensburg

**Project:** Implementation and Optimization of Circular RNA Analysis Bioinformatics Pipeline: Pre-processing, Alignment, Detection, Differential Expression and Functional Annotation.

Performed bioinformatics analysis on circRNAs using backsplice region identification. Developed and optimized a computational protocol for detecting and quantifying circRNAs from RNA-Seq data. Investigated the differential expression of circRNAs and their relationship with linear transcripts, identifying significant differences between B-Cell and Monocyte cell samples.

### • Research Internship at MUN, Canada (MITACS Globalink Scholarship)

(May'21-Aug'21)

*Project, Report*

**Supervisor:** [Dr. Sevtap Savas](#), Department of Genetics, Memorial University of Newfoundland, St. Johns.

**Project:** Gene Expression Profiles, Biological Pathways, and Disease Characteristics associated with BRM Expression Levels in Lung Adenocarcinoma Tumors.

Examined the correlation between BRM (SMARCA2) expression levels in LUAD tumors and the expression profiles of other genes, their pathways, and patient survival rates using TCGA LUAD dataset. Conducted QC, Mann-Whitney U Test, Bonferroni Correction, GO/Reactome Pathway Analysis, and Survival Analysis utilizing DAVID's Functional Annotation Clustering tool.

### • Research Internship at IIT-Roorkee, India (SPARK Fellowship)

(April'20–July'20)

*Project*

**Supervisor:** [Dr. Prateek K. Jha](#), Associate Professor, Department of Chemical Engineering, IIT-Roorkee.

**Project:** Optimization of Polymer Predictors for Drug Delivery Systems, using Machine Learning Approaches.

Conducted a study to optimize polymer selection for specific drugs using Machine Learning techniques in RStudio. Data were collected on 102 polymers and 9 descriptors were used to perform Exploratory Data Analysis, followed by pre-processing through outlier removal and feature selection. A multivariate linear regression model was chosen as the most accurate for prediction purposes.

## PUBLICATIONS

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1. **Negi, S**, Stenton SL, Berger SI, Canigiula P, McNulty B, Violich I, Gardner J, Hillaker T, O'Rourke SM, O'Leary MC, Carbonell E, Austin-Tse C, Lemire G, Serrano J, Mangilog B, VanNoy G, Kolmogorov M, Vilain E, O'Donnell-Luria A, Délot E, Miga KH, Monlong J, Paten B. **Advancing long-read nanopore genome assembly and accurate variant calling for rare disease detection**. Am J Hum Genet. 2025 Jan 22:S0002-9297(25)00002-3. doi: 10.1016/j.ajhg.2025.01.002.
2. Monlong, J., Chen, X., Barseghyan, H., Rowell, W.J., **Negi, S.**, Nokoff, N., Mohnach, L., Hirsch, J., Finlayson, C., Keegan, C.E., Almalvez, M., Berger, S.I., de Dios, I., McNulty, B., Robertson, A., Miga, K.H., Speiser, P.W., Paten, B., Vilain, E., Délot, E.C (2025). **Long-read sequencing resolves the clinically relevant CYP21A2 locus, supporting a new clinical test for Congenital Adrenal Hyperplasia**. medRxiv. 2025.02.07.25321404. doi: 10.1101/2025.02.07.25321404.
3. Billingsley, K.J., Meredith, M., Daida, K., Alvarez Jerez, P., **Negi, S.**, Malik, L., Genner, R.M., Moller, A., Zheng, X., Gibson, S.B., Mastoras, M., Baker, B., Kouam, C., Paquette, K., Jarreau, P., Makarious, M.B., Moore, A., Hong, S., Vitale, D., Shah, S., Monlong, J., Pantazis, C.B., Asri, M., Shafin, K., Carnevali, P., Marengo, S., Auluck, P., Mandal, A., Miga, K.H., Rhie, A., Reed, X., Ding, J., Cookson, M.R., Nalls, M., Singleton, A., Miller, D.E., Chaisson, M., Timp, W., Gibbs, J.R., Phillippy, A.M., Kolmogorov, M., Jain, M., Sedlazeck, F.J., Paten, B., Blauwendraat, C (2024). **Long-read sequencing of hundreds of diverse brains provides insight into the impact of structural variation on gene expression and DNA methylation**. bioRxiv. 2024.12.16.628723. doi: 10.1101/2024.12.16.628723.
4. **Negi, S.**, Chaudhuri, A., Kumar, D.N. et al. **Nanotherapeutics in autophagy: a paradigm shift in cancer treatment**. Drug Deliv. and Transl. Res. 12, 2589–2612 (2022). doi: 10.1007/s13346-022-01125-6
5. Tiwari JK, **Negi, S.**, Kashyap M, Nizamuddin S, Singh A and Khattri A (2022). **Pan-Cancer Analysis Shows Enrichment of Macrophages, Overexpression of Checkpoint Molecules, Inhibitory Cytokines, and Immune Exhaustion Signatures in EMT-High Tumors**. Front. Oncol. 11:793881. doi: 10.3389/fonc.2021.793881

## TALKS AND PRESENTATIONS

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1. Invited talk at **Chan Zuckerberg Initiative**, Redwood City. (2024). Long-read nanopore sequencing for advancing rare disease diagnosis.
2. Lightning talk and poster presentation at the **HPRC conference**, Santa Cruz (2024). Cost-efficient long-read nanopore sequencing for enabling rare disease diagnosis.
3. Poster presentation at **Advances in Genome Biology and Technology, Precision Health (AGBTph)** (2023). Advancing long-read nanopore genome assembly and accurate variant calling for rare disease detection.
4. Poster presentation at the **Canadian Cancer Research Alliance (CCRA) Conference**, Toronto (2021). Gene expression profiles, biological pathways, and disease characteristics associated with BRM expression levels in lung adenocarcinoma tumors.

## HONORS AND ACADEMIC ACHIEVEMENTS

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- **IIT-BHU President's Gold Medalist' 2022** - For outstanding performance in academics among all disciplines of B.Tech. Examination 2022
- Felicitation and media coverage as the President's Gold Medalist at IIT-BHU, with interviews by several newspapers, including **Times of India**.
- **IIT-BHU Varanasi Medal, Aruna and Malviya Medal, Prof. Gopal Tripathi Memorial Gold Medal & Shri J.N. Kapoor Gold Medal** - For standing First at the B.Tech. in Pharmaceutical Engineering & Technology Examination, 2022
- **Late Dr. R.N. Singh and Mrs. Uma Singh Medal** - For securing the highest CPI among the girl students at the B.Tech. Examination, 2022
- **Late Shri Shyam Sunder Lal Razdan Memorial Gold Medal & Shri Raj Kishore Kapoor Silver Medal** - For securing the highest percentage of marks in B.Tech. Examination, 2022
- **Late Sundari Devi Gold Medal** - For securing the highest CPI > 8.50 as a girl student in Pharmaceutical Engineering and Technology at the B.Tech. Examination, 2022
- **Smt. Arati Paul and Prof. Binod Bihari Paul Gold Medal & Umesh Pratap Singh Gold Medal** - For securing the highest marks in IV Year Examination among all the students of B.Tech. Examination, 2022
- **MITACS Globalink Scholar, Canada** - Selected amongst 300 students from all over the World.
- **DAAD-WISE Scholar, Germany** - Selected amongst 150 students from all over India.
- **SPARK, IIT-Roorkee Research Fellowship** - Selected amongst 50 students all over India.
- **Best Presentation Award** - For the best presentation of my research project at SPARK, IIT-Roorkee.
- **IBGAA GSAS Awardee** - Awarded the Graduate Study Application Scholarship by the IIT-BHU Global Alumni Association for extraordinary academic excellence.
- JEE (Joint Entrance Examination) Advanced (2018) **All-India Rank – 14167** amongst 1.6 million candidates.