

Shunhua Han

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EDUCATION **University of Georgia**, Athens, GA, USA
Ph.D. in Bioinformatics, December 2021

- Dissertation Title: “Novel computational strategies for the analysis of transposable elements in *Drosophila* cell culture genomes”
- Advisor: Casey Bergman

East China University of Science and Technology, Shanghai, China
B.S. in Pharmaceutical Sciences, June 2015

PROFESSIONAL EXPERIENCE **Illumina**, San Diego, CA, USA
Sr Staff Bioinformatics Scientist **Nov 2025 - present**

Resolve challenging and disease causing genes using constellation mapped reads and standard whole genome sequencing

Staff Bioinformatics Scientist **Nov 2023 - Oct 2025**

- Improved variant calling accuracy in challenging and disease causing genes, notably in *PMS2*, significantly impacting Lynch syndrome diagnostics.
- Research blog published at www.illumina.com/science/genomics-research/articles/PMS2-small-variant-detection.html

Senior Bioinformatics Scientist **Jan 2022 - Oct 2023**

- Led the research and development of *HBA1/2* copy number genotyping method for diagnosis and carrier screening of alpha thalassemia, which is one of the most common monogenic diseases that has 5% global carrier frequency.
- Research blog published at www.illumina.com/science/genomics-research/articles/HBA-targeted-caller.html

ACADEMIC EXPERIENCE **University of Georgia**, Athens, GA, USA
Graduate Student **August 2016 - December 2021**

Developed software that detects TEs in long read sequencing data (PacBio/Nanopore)

- Developed a novel pipeline for detecting transposable elements (TE) from long read sequencing data with high efficiency and accuracy.
- The program is open sourced and released in github.com/bergmanlab/TELR.

Developed a protocol for *Drosophila* cell line authentication

- Developed novel approach for identifying transposable element (TE) insertions using WGS data. (github.com/bergmanlab/ngs_te_mapper2).
- Developed protocol to authenticate *Drosophila* cell lines based on their unique TE profiles.
- Report first misidentification events in *Drosophila* cell lines involving mbn2 and Sg4.

Detected patterns of transposable element (TE) insertion activity among *D. melanogaster* S2 cell lines

- Genomic analysis of 32 whole genome sequencing data of *D. melanogaster* S2 cell lines including CNV calling and TE prediction.
- Phylogenetic analysis including tree building and ancestral state reconstruction revealed ongoing transposition pattern of TEs in S2 cell lines.
- Developed and implemented strategy to estimate TE normalized coverage using next generation sequencing data.

Investigated *P* element target site preference using machine learning

- Engineered 30 features including tissue-specific gene expression data and chromatin activity data for 15860 target sites.
- Identified one specific group of target site that was suggested to be the set that *P* element will avoid even with saturating mutagenesis, and another negative group that was suggested to be potential *P* element target in the future.

Teaching Assistant

Fall, 2020

- Graduate level course “Applied Genome Analysis”
- Gave guest lecture “Introduction to Bioconductor and ggplot” at a graduate level course “Applied Genome Analysis”.
- Duties also included office hours, lab course assistance, and assignment grading.

SELECTED PUBLICATIONS

Han, S., Dias, G.B., Basting, P.J., Viswanatha, R., Perrimon, N. and Bergman, C.M., 2022. Local assembly of long reads enables phylogenomics of transposable elements in a polyploid cell line. *Nucleic Acids Research*, 50(21), pp.e124-e124.

Han, S., Dias, G.B., Basting, P.J., Nelson, M.G., Patel, S., Marzo, M. and Bergman, C.M., 2022. Ongoing transposition in cell culture reveals the phylogeny of diverse *Drosophila* S2 sublines. *Genetics*, 221(3), p.iyac077.

Han, S., Basting, P.J., Dias, G.B., Luhur, A., Zelfhof, A.C. and Bergman, C.M., 2021. Transposable element profiles reveal cell line identity and loss of heterozygosity in *Drosophila* cell culture. *Genetics*, 219(2), p.iyab113.

PATENTS

Han, Shunhua, Vitor Ferreira Onuchic, Xiao Chen. Methods and systems for determining copy number variant genotypes. *WO Patent Application No. WO2024010812A2 (PCT/US2023/026935)*, Publication date: January 11, 2024.

Vitor Ferreira Onuchic, **Han, Shunhua**. Region-ambiguous variant detection. *US Patent Application No. 18/674,538*, Publication date: November 28, 2024.

Han, Shunhua, Daniel Andrews, Vitor Ferreira Onuchic. Multi-region joint detection. *US Patent Application No. 18/674,279*, Publication date: November 28, 2024.

SELECTED
PRESENTATIONS

A Rapid, Novel Approach to Rare Disease and Clinical Genetic Variant Discovery using On-flowcell Proximity Sequencing and Haplotype-resolved Variant Calling.

Platform presentation at 2026 ACMG Clinical Genetics Meeting, Mar 12, Baltimore, MD.

Haplotype-resolved variant detection in medically relevant and paralogous genes using multi-region joint detection with Illumina on-flowcell proximity sequencing.

Poster presentation at 2025 Advances in Genome Biology and Technology (AGBT) Precision Health, Sep 9, San Diego, CA.

Overcoming the challenges of variant calling in *PMS2* high homology regions for improved lynch syndrome diagnosis using whole-genome sequencing.

Poster presentation at 2023 Intelligent Systems For Molecular Biology (ISMB) Conference, Jul 25, Lyon, France.

Targeted detection of clinically relevant variants in *HBA1/2* from whole-genome sequencing data.

Poster presentation at 2022 Advances in Genome Biology and Technology (AGBT) Precision Health Meeting, Sep 8, San Diego, CA.

Genomic insights into the pattern of transposable element proliferation in *Drosophila* S2 cells.

Invited Talk at 2018 Mobile Genetic Elements and Genome Plasticity, Feb 11 - 15, Santa Fe, NM.

TECHNICAL SKILLS

Python, R, C++, Bash.

Pipelines/Infra: Nextflow, Docker, AWS

Experienced with computational methods for genomic variant calling, particularly in complex and repetitive regions.

OTHER
PUBLICATIONS

De La Vega, F.M., Irvine, S.A., Anur, P., Potts, K., Kraft, L., Torres, R., Kang, P., Truong, S., Lee, Y., **Han, S.** and Onuchic, V., 2025. Benchmarking of germline copy number variant callers from whole genome sequencing data for clinical applications. *Bioinformatics Advances*, 5(1), p.vbaf071.

Chen, J., Basting, P.J., **Han, S.**, Garfinkel, D.J., and Bergman, C.M., 2023. Reproducible evaluation of transposable element detectors with McClintock 2 guides accurate inference of Ty insertion patterns in yeast. *Mobile DNA*, 14(8).

Mariyappa, D., Rusch, D.B., **Han, S.**, Luhur, A., Overton, D., Miller, D.F., Bergman, C.M. and Zelfhof, A.C., 2022. A novel transposable element-based authentication protocol for *Drosophila* cell lines. *G3*, 12(2), p.jkab403.

Han, S., 2021. Novel computational strategies for the analysis of transposable elements in *Drosophila* cell culture genomes (Doctoral dissertation, University of Georgia).

Bergman, C.M., **Han, S.**, Nelson, M.G., Bondarenko, V. and Kozeretska, I., 2017. Genomic analysis of P elements in natural populations of *Drosophila melanogaster*. *PeerJ*, 5, p.e3824.

OTHER
PRESENTATIONS

Haplotype-resolved variant detection in medically relevant and paralogous genes using multi-region joint detection with Illumina constellation mapped reads.

Poster presentation at 2025 American Society of Human Genetics (ASHG), Oct 17, Boston, MA.

A rapid, novel WGS approach detects pathogenic variants in high homology pseudogenes using on-flowcell proximity sequencing and haplotype-resolved variant calling.

Poster presentation at 2025 American Society of Human Genetics (ASHG), Oct 17, Boston, MA.

Predicting *P* element target site preferences using machine learning.
Invited Talk at 2019 Advancing Informatics in Government and Industry symposium, Jul 25, Athens, GA.

Genomic insights into the pattern of transposable element proliferation in *Drosophila* S2 cells.
Poster presentation at 2017 Mobile Genetic Elements Conference, Aug 29 - 31, Woods Hole, MA.

HONORS AND
AWARDS

University of Georgia Graduate Student Travel Award, 2017-2019