

Sanjar Hudaiberdiev, PhD

Staff Scientist, NCBI/NIH

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Scientific research areas

Genomics, Gene Regulation, Disease Genetics (Type 2 Diabetes)

Evolution, Origin of Life

Machine/Deep Learning, Statistics

Technical Skills & Domain Knowledges

Biology	Gene regulation (enhancers, silencers), 3D chromatin organization, condensates Type 2 Diabetes: pancreatic islet (β) cells, islet-specific TFs, risk loci (GWAS) PopGen: common/rare variant analysis, LD blocks, GWAS, PRS <u>Evolution</u> : non-coding regions, ultra-conserved regions, human-specific evolution (HARs, hCONDELs), protein(domain) evolution, phylogenetics/phylogenomics genomic synteny, host-parasite arms race
ML/stats	Statistical Inference, Biostatistics Bayesian modeling, Probabilistic Reasoning, Causality ConvNets, LSTMs, Transformers
ML tools	Tensorflow, PyTorch, Keras Scikit-learn, NumPy/SciPy, R, MATLAB/Octave
Bioinformatics	GWAS, fine-mapping, e/s/raQTLs, MPRA ChIP-seq, RNA-seq, ATAC-seq, Hi-C ENCODE, Roadmap, HCA, 4D Nucleome, UK Biobank Containers (Docker, Singularity), Workflows (Nextflow, Snakemake) Massive data processing, parallel computing, HPC (SLURM, SGE)
Soft. Dev.	OOP, functional programming, Unit tests, Git, CI/CD
Languages	Actively using: Python, R, Rust (learning), Bash Have used: MATLAB, C#, C/C++, Perl
Databases	MySQL, PostgreSQL, SQLite, SQL Server 2008/2010, Oracle 10g.
OS	Linux, MacOS

Work Experience

2023 –	Staff Scientist. NCBI, NIH, Bethesda, MD - Integrating multi-omics (TF/Histone ChIP-seq, DHS, ATAC-seq, HiC, RNA-binding proteins' binding sites, methylome) data to model disease transcriptional codes using multi-modal fusion deep neural networks to detect disease-causal genetic variation. - Using Deep Learning to assess the phenotypic impacts of human-specific (vs. Macaques and Chimpanzees) INDELs in enhancers.
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- 2020 – 2023 | Research Fellow. NCBI, NIH, Bethesda, MD
- Developed TREDNet: A novel Deep Learning method for enhancer detection.
 - Lead a large multidisciplinary collaboration project to apply TREDNet to detect GWAS candidate causal mutations linked to Type 2 Diabetes and experimentally verified.
 - Developed a model for explaining the high-occupancy target (HOT) regions in humans using transcriptional condensates.
- 2018 – 2020 | Postdoctoral Fellow. NCBI, NIH, Bethesda, MD
- Mentor: Ivan Ovcharenko, PhD*
- Spearheaded the transition to Deep Learning methods to study the enhancers.
 - Automated the distributed training of large models (Keras+Tensorflow) on Google Cloud and Biowulf (NIH's HPC cluster).
 - Applied ConvNets and LSTMs to model gene regulation and decompose LD blocks.
 - Managed and implemented the deployment of decades-worth codebase developed in-house to containers (Docker, singularity) and workflows management systems (Nextflow, Snakemake).
- 2015 – 2018 | Postdoctoral Fellow. NCBI, NIH, Bethesda, MD
- Mentor: Eugene Koonin, PhD*
- Studied the general evolution of CRISPR/Cas systems, as well as deep focus on protein CRISPR-associated 4 (Cas4). Paper is cited >55 times, including by Prof. Jennifer Doudna, and has given me an Erdős number of 3.
 - Analyzed evolutionary links between microbial adaptive immune systems and other selfish elements.
 - Gained an expert understanding of microbial genetics, comparative genomics, molecular evolutionary analysis.
- 2011 – 2014 | Graduate research program ICGEB, Trieste, Italy
- **Thesis project:** Functional annotation of *Quorum Sensing* systems in bacteria using subsystem based approach.
 - Implemented a wide range of algorithms and tools of computational genomics and sequence analysis.
- 2010 – 2011 | Software developer, Octopus, Bishkek, Kyrgyzstan
- Developed a concept for data analysis module for an existing commercial software for micro-financing.
 - Implemented financial data analysis module in .NET environment using C# and MS SQL Server.
 - Developed an ETL module to consolidate the data from government and third-party vendors using Pentaho and Python.
- 2009 – 2010 | Oracle Database Developer, Demirbank, Bishkek, Kyrgyzstan
- Designed and implemented backup strategies of Oracle database and decreased the backup time by a factor of 10.
 - Lead the project of transitioning the existing infrastructure to Oracle Blade Modules.
 - Conducted SQL tuning and optimization which lead to reduction in ATM transaction latencies during the peak times from 10 seconds to < 1 second.
- 2008 - 2009 | Research assistant, Istanbul Technical University, Turkey
- Mentor: Prof. Zehra CATALTEPE.*
- Designed and implemented Machine Learning experiments on protein function prediction using profile-HMMs.
 - Obtained a formal graduate training on classical Machine Learning (PRML by Christopher Bishop)

Education

- 09/2011 - 09/2014 PhD in Bioinformatics, **ICGEB**, Trieste, Italy
Thesis: "Computational analysis of quorum sensing systems in bacterial genomes: Developing automated annotation tools."
Supervisor: Prof. Sándor PONGOR
- 09/2006 - 09/2008 MSc in Machine Learning, **Istanbul Technical University**, Istanbul, Turkey
- 09/2002 - 09/2006 BSc in Computer Engineering, **Erciyes University**, Kayseri, Turkey

Publications

C. Hill, **S. Hudaiberdiev**, I. Ovcharenko

ChromDL: A Next-Generation Regulatory DNA Classifier ISMB/ECCB 2023 (in submission)
bioRxiv (<https://doi.org/10.1101/2023.01.27.525971>)

S. Hudaiberdiev, I Ovcharenko

Sequence characteristics and an accurate model of abundant hyperactive loci in the human genome
eLife 2023 (in submission)
bioRxiv (<https://doi.org/10.1101/2023.02.05.527203>)

S. Hudaiberdiev, D. Taylor, W Song, N Narisu, RM. Bhuiyan, H. Taylor, T Yan, A Swift, L Bonnycastle, DIAMANTE Consortium, M Stitzel, M Erdos, I Ovcharenko, F. S. Collins

Modeling islet enhancers using deep learning identifies candidate causal variants at loci associated with T2D and glycemic traits *PNAS*. 2023 (being reviewed)
medRxiv (<https://doi.org/10.1101/2022.05.13.22275035>)

EZ Kvon, Y Zhu, G Kelman, CS Novak, I Plajzer-Frick, M Kato, T H Garvin, Q Pham, AN Harrington, R Hunter, J Godoy, E Meky, J Akiyama, V Afzal, S Tran, F Escande, B Gilbert-Dussardier, N Jean-Marçais, **S Hudaiberdiev**, I Ovcharenko, MB Dobbs, CA Gurnett, S Manouvrier-Hanu, F Petit, A Visel, DE Dickel, LA Pennacchio

Comprehensive in vivo interrogation reveals phenotypic impact of human enhancer variants. *Cell*. 2020

Hudaiberdiev S., Shmakov S, Wolf YI, Terns M, Makarova KS and Koonin EV **2017**.

Phylogenomics of Cas4 family nucleases. BMC Evol. Biol. 2017 PMID: 29179671

Hudaiberdiev, S., Choudhary, K.S., Vera, R., Gelencser, Z., Lamba, D. and Pongor, S. **2015**.

Census of solo luxR genes in bacteria. Frontiers in Cell. Infect. Microbiol. 2015. doi:10.3389

Dogsa, I; Choudhary K.S; Marsetic, Z; **Hudaiberdiev, S**; Vera, R; Pongor, S, Mandic-Mule, I **2014**.
ComQXPA quorum sensing systems may not be unique to Bacillus subtilis: a census in prokaryotic genomes. PLoS One. 9(5):e96122. doi: 10.1371

Choudhary, K.S.; **Hudaiberdiev, S.**; Gelencsér, Z.; Coutinho, B.G.; Venturi, V.; Pongor., S. **2013**.

The organization of the Quorum Sensing luxI/R Family Genes in Burkholderia. Int J Mol Sci. 13727

Gelencsér, Z.; Choudhary, K.S.; Coutinho, B.G.; **Hudaiberdiev, S.**; Galbáts, B.; Venturi, V.; Pongor, S. **2012** *Classifying the topology of AHL-driven quorum sensing circuits in proteobacterial genomes.* *Sensors.* 12(5), 5432-5444.

Gelencsér, Z.; Galbáts, B.; Gonzalez, J.F.; Choudhary, K.S.; **Hudaiberdiev, S.**; Venturi, V.; Pongor, S. **2012.** Chromosomal arrangement of AHL-driven quorum sensing circuits in *Pseudomonas*. *ISRN Microbiology.* 2012, 6.

Oral presentations

- 2023 *Sequence characteristics and an accurate model of abundant hyperactive loci in the human genome* NLM IRP Seminar
- 2022 *Transcriptional regulation and disease genetics using Deep Learning* NLM IRP Seminar
- 2021 *Deep causality in non-coding GWAS SNPs.* NLM Summer Lectures Series. NLM
- 2020 *Using Deep Learning to infer causality of GWAS SNPs.* METU, Ankara, Turkey
- 2019 *Recurrent Language model for enhancers.* NLM Summer Lectures Series. NLM
- 2019 *Understanding the grammar of enhancers using Deep Learning.* NHGRI
- 2019 *Causal mutations in ZRS enhancer region using Deep Learning.* LBNL, Berkeley
- 2017 *Phylogenomics of Cas4 family nucleases.* AiChE. Raleigh, NC
- 2016 *Current topics in biological sequence analysis.* ICGEB. Trieste, Italy

Posters

- 2021 *Hyperactive loci within non-coding regulatory regions of human genome*
Mechanisms of Eukaryotic Transcription, CSHL, NY.
- 2019 *Two-stage Deep Learning cross-tissue predictor of enhancers shows unprecedented accuracy in detection of causal regulatory variants.*
Mechanisms of Eukaryotic Transcription, CSHL, NY.
- 2019 *Using Deep Learning to understand the grammar of enhancers.*
RECOMB, Washington DC
- 2017 *Phylogenomics of Cas4 family nucleases.* *International conference on CRISPR Technologies*, Raleigh, NC
- 2013 *Computational approaches to microbial communication/quorum sensing signaling.*
BAGECO, Ljubljana, Slovenia

Awards

- Fellows Award for Research Excellence 2019, NIH, Bethesda, MD
- ICGEB PhD fellowship 2011, ICGEB, Trieste, Italy
- Scholarship for research in Machine Learning/Bioinformatics. 2008, TUBITAK, Istanbul, Turkey
- Scholarship for BS final year project. 2007, TUBITAK, Kayseri, Turkey
- Scholarship of Turkish ministry of education for undergraduate studies. 2002/2007, Turkey
- Bronze medals in national physics olympiads for highschool students. 2001/2002, Kyrgyzstan

Teaching & Mentorship

- Co-mentor, Post-bac student, 2020 NIH
- Co-mentor, Summer research student, 2019 NIH
- Lead instructor, Summer intern journal club, 2018 NIH
- Co-mentor, Summer research student, 2018 NIH

Peer-reviewing for Scientific Journals

BMC Bioinformatics, NAR Database, Frontiers in Microbiology

Organization & Leadership

Committee

2023 Fellow Award for Research Excellence. Committee member

2022 Fellow Award for Research Excellence. Committee member

Organised

2013 Bioinformatics: Computer Methods in Molecular Biology –ICGEB, Trieste, Italy

2014 Bioinformatics: Computer Methods in Molecular Biology –ICGEB, Trieste, Italy

Languages

Kyrgyz	Native
English, Turkish	Fluent
Italian, Russian	Advanced