SINEM DEMIRKAYA BUDAK

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Work Experience

• 01.2023 - 09.2024

Bioinformatics Engineer, Velsera Inc., Ankara, TURKEY (Remote)

- Developed, optimized and prepared for releasing a novel Family Trio Analysis Workflow for de novo mutation and loss of heterozygosity (LoH) detection.
- Evaluated performance and created reports and visualizations to simplify the interpretation of analysis results.
- 01.2020 01.2023

Bioinformatics Analyst, Seven Bridges Genomics Inc., Ankara, TURKEY (Remote)

- Optimized and benchmarked various bioinformatics tools and workflows on Seven Bridges Platform and other platforms.
- Took role in bioinformatics tasks of data QC, preprocessing, genome alignment and variant calling, postprocessing.
- Containerized and enhanced the performance of multiple bioinformatics tools on Seven Bridges Platform and Cancer Genomics Cloud.
- Led the challenge for Seven Bridges and we won the best performance for accurately calling variants in the MHC region of Illumina short reads in the precisionFDA Truth Challenge V2.
- Developed a novel de novo mutation detection workflow in collaboration with an external partner.
- 05.2018 01.2020

Assistant Project Manager and Lab Specialist, Seven Bridges Genomics Inc., Ankara, TURKEY

- Performed next generation sequencing (NGS) in a national genome project with Illumina NovaSeq 6000
 System.
- Performed all laboratory processes for NGS like DNA isolation and library preparation, trained and oriented clients about all protocols from blood sample retrieval to NGS data generation.
- Took active role in optimizing and benchmarking various bioinformatics tools and workflows on Seven Bridges Platform.
- 01.2016 04.2018

Teaching & Research Assistant, TOBB University of Economics and Technology (TOBB ETU), Ankara, TURKEY

- Studied effects of several SNPs on an eye disease utilizing DNA isolation techniques (salt-out and using commercial kit) from several sample types including whole blood, PCR/RFLP, real-time PCR and ELISA methods for investigating isolated DNA.
- Researched, ordered and installed lab equipment to a central lab and guided graduate students for using these equipment.
- Prepared and submitted research projects, consent, ethics and phenotypic forms for projects including human subjects.
- o Taught Molecular Biology and Genetics laboratory and Physics 101 laboratory classes.
- 07.2013 09.2013

Intern, Institute of Systematic Botany and Mycology, University of Munich (LMU München), Munich, GERMANY

- Studied three species of Caricaceae for validating their phylogenetic relationships by utilizing chromosome counting technique (I was acknowledged for my contribution in this publication: http://www.springer.com/br/book/9783658102661).
- Helpeds lab technicians with their regular workflow in several studies including sample collection, sample processing for chromosome counting, PCR, agarose gel electrophoresis, and chromosome counting with DAPI staining under fluorescent microscope.

10.2014 - 06.2015, 10.2012 - 01.2013

Special Tutor, http://odevodtu.com, Ankara, TURKEY

Tutored primary and secondary education students in math and science classes.

Education

• 09.2018 - 01.2024

PhD, Medical Informatics, Middle East Technical University (METU)

CGPA: 3.75 / 4.00

- Took PhD courses including programming for informatics, biological databases and data analysis tools,
 OOP and data structures, structural bioinformatics, probabilistic and statistical modeling for
 bioinformatics, protein and DNA sequence analysis, fundamentals of mathematics for information
 systems, medical informatics, research methods and statistics for cognitive science.
- Studied and passed PhD qualifiers.
- Withdrew from the PhD program during the thesis design phase because of time limitations.
- 01.2016 04.2018

Master's Degree, Biomedical Engineering, TOBB University of Economics and Technology (TOBB ETU) CGPA: 4.00 / 4.00

- Investigation of the relationship between clusterin genetic polymorphisms and clusterin level of tear and aqueous humor in pseudoexfoliation syndrome and glaucoma (Master thesis study).
- 09.2008 06.2015

BSc, Molecular Biology and Genetics, Middle East Technical University (METU)

CGPA: 2.57 / 4.00

- Spring 2014 Laboratory practices of green microalgae (Chlamydomonas reinhardtii) to produce biodiesel from microalgae.
 - Middle East Technical University (METU), Ankara, TURKEY
- Fall 2013 Physiological and biochemical response to boron toxicity in two barley cultivars (Zafer 160 and Tarm 92). To study the effects of boron toxicity; growth of root and shoot, antioxidant defense mechanism (Ascorbate peroxidase, Catalase, Glutathione reductase), relative water content, proline content, H2O2 content, malondialdehyde content, protein concentration and the pigment concentration are investigated.
 Middle East Technical University (METU), Ankara, TURKEY
- Summer 2013 Chromosome counting for three species of Caricaceae C.parviflora, J. caudata, J. heterophylla by using classical cytogenetic techniques and DAPI staining.
 Institute of Systematic Botany and Mycology, University of Munich (LMU München), Munich, GERMANY
- 11.2011 06.2015

BSc, Business Administration (Open Education), Anadolu University

CGPA: 2.65 / 4.00

• 09.2004 - 06.2008

Mathematics and Science, Mehmet Akif Ersoy High School

Diploma grade: 93.78 / 100

<u>Publications</u>

- Shadrina M, Kalay Ö, Demirkaya-Budak S, LeDuc CA, Chung WK, Turgut D, Budak G, Arslan E, Semenyuk V,
 Davis-Dusenbery B, Seidman CE, Yost HJ, Jain A, Gelb BD. Efficient identification of de novo mutations in family
 trios: a consensus-based informatic approach. *Life Science Alliance*. 2025 Jun 1.
 https://www.life-science-alliance.org/content/8/6/e202403039
- Arslan E, Turgut D, Kalay Ö, Demirkaya-Budak S, Budak G, Jain A. Pangenome References Improve Biomarker Estimation from Tumor Sequencing Data. bioRxiv. 2024 Nov 15:2024-11. https://doi.org/10.1101/2024.11.14.623554

- Tetikol H S, Turgut D, ..., Demirkaya-Budak S, ..., Davis-Dusenbery B N. 2022. Pan-African genome demonstrates how population-specific genome graphs improve high-throughput sequencing data analysis. *Nature Communications*, 13(1), 4384.
 - https://www.nature.com/articles/s41467-022-31724-3
- Olson N D, Wagner J, McDaniel J, ..., Demirkaya-Budak S, ..., Zook J M. 2022. PrecisionFDA Truth Challenge V2: Calling variants from short and long reads in difficult-to-map regions. *Cell genomics*, 2(5). https://www.sciencedirect.com/science/article/pii/S2666979X22000581
- Can Demirdöğen B, Demirkaya-Budak S, Özge G, Mumcuoğlu T. 2019. Evaluation of Tear Fluid and Aqueous Humor Concentration of Clusterin as Biomarkers for Early Diagnosis of Pseudoexfoliation Syndrome and Pseudoexfoliative Glaucoma. *Current Eye Research*. https://doi.org/10.1080/02713683.2019.1698055
- Can Demirdöğen B, Koçan Akçin C, Göksoy E, Yakar G, Öztepe T, Demirkaya-Budak S, Oflaz S. 2019. Paraoxonase 1 (PON1) promoter (-107T/C) and coding region (192Q/R and 55L/M) genetic variations in pseudoexfoliation syndrome and pseudoexfoliative glaucoma risk. *Graefe's Archive for Clinical and Experimental Ophthalmology*. https://doi.org/10.1007/s00417-019-04408-w

<u>Skills</u>

- Programming: Python, JavaScript, R, HTML, MySQL, PostgreSQL, SQLite
- Technologies & Libraries: Docker, CWL, Git, BioPython, pysam, Matplotlib, Pandas, Numpy, Scipy, Scikit-learn
- Bioinformatics tools: Data analysis (BWA, GATK, STAR, Rabix, IGV, Cytoscape, Autodock4, PyMol, UCSC Chimera, Ensembl API), NGS data QC (FastQC, FastQ Screen, Cutadapt, Atropos, Fastp), data manipulation (SAMTools, BCFTools)
- Software/OSes: RStudio, MS Office tools, Minitab, SPSS, MS Windows, Linux, macOS
- Project management: Jira, Kanban, Agile, Confluence
- Laboratory: NGS, Light/Fluorescence Microscope, PCR/RFLP, Electrophoresis, Spectrophotometry/Fluorometer, Library preparation, Real-Time PCR, DNA isolation from several sample types w/ or w/o an automated system, ELISA

Awards, Certificates & Courses

- Best performance for accurately calling variants in the MHC region of Illumina short reads in the precisionFDA
 Truth Challenge V2 U.S. Food and Drug Administration, Jun 2020
 - I led the challenge for Seven Bridges and we won the best performance for accurately calling variants in the MHC region of Illumina short reads in the precisionFDA Truth Challenge V2
- Anadolu University Certificate of Honor: 2013 2014 Spring Semester
- METU Certificate of High Honor: 2010 2011 Fall Semester
- Website Design: BELTEK Technical Education Course of Gazi University/Ankara Metropolitan Municipality 12.04.2010 – 06.06.2010 (72 hours)

References

Amit Jain – Graph Technology Director

Velsera Inc., London, England, United Kingdom

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Prof. Dr. Birsen Can Demirdöğen

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