

## SİNEM DEMİRKAYA BUDAK

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[sinemdemirkayabudak.github.io](https://sinemdemirkayabudak.github.io)

### **Work Experience**

- 01.2023 – 09.2024

#### **Bioinformatics Engineer, Velsara Inc., Ankara, TURKEY**

- 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**

- 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.
- 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>).
- Helps 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://odevodu.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, H<sub>2</sub>O<sub>2</sub> 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

## **Publications**

- 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. Automated Identification of Germline de novo Mutations in Family Trios: A Consensus-Based Informatic Approach. *bioRxiv [Preprint]*. 2024 Mar 13.  
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC10979888/>
- 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>

### **Oral Presentations**

- **Demirkaya S**, Can Demirdöğen B, Özge G, Mumcuoğlu T. Relationship between clusterin polymorphisms and pseudoexfoliation syndrome and glaucoma. In vitro Diagnostic Biomarkers Symposium. May 10-12, 2017. İzmir, Turkey (Oral presentation). *Turkish Journal of Biochemistry*. 2017; vol 42(S2): pp 52, S-12.
- **Demirkaya S**, Can Demirdöğen B, Özge G, Mumcuoğlu T. Analysis of the relation between clusterin rs11136000 polymorphism and pseudoexfoliation syndrome. TBD Biochemistry Days. November 2–5, 2016. Sivas, Turkey (Oral presentation). *Turkish Journal of Biochemistry*. 2016; vol 41 (S3), S036.

### **Poster Presentations**

- **Demirkaya Budak S**, Can Demirdöğen B, Özge G, Mumcuoğlu T. The association between clusterin rs11136000 C/T single nucleotide polymorphism and clusterin protein level in tear fluid in pseudoexfoliative glaucoma risk. The 43rd FEBS Congress. July 7-12, 2018. Prague, Czech Republic. *FEBS Open Bio*. 2018; vol. 8 (S1): pp 255, P.09-147-Wed. doi:10.1002/2211-5463.124534
- **Demirkaya-Budak S**, Can Demirdöğen B, Özge G, Mumcuoğlu T. The Association between clusterin rs11136000 C/T single nucleotide polymorphism and pseudoexfoliative glaucoma risk. International Eurasian Conference on Biological and Chemical Sciences. April 26-27, 2018. Ankara, Turkey. *Eurasian Journal of Biological and Chemical Sciences*, Abstract Book 2018; pp 584.
- **Demirkaya Budak S**, Can Demirdöğen B, Özge G, Mumcuoğlu T. Evaluation of the effect of clusterin rs2279590 single nucleotide polymorphism in pseudoexfoliation syndrome risk and on clusterin protein level in aqueous humor. The 42nd FEBS Congress. September 10–14, 2017. Jerusalem, Israel. *The FEBS Journal*. 2017; vol. 284 (S1), pp 138, P.1.5-054. DOI: 10.1111/febs.14174
- Can Demirdöğen B, Yakar G, Göksoy E, **Demirkaya S**, Koçan C, Özge G, Mumcuoğlu T. Relationship between paraoxonase 1 (PON1) promoter (-107T/C) and coding region polymorphisms (192Q/R and 55L/M) and Pseudoexfoliation syndrome. The 42nd FEBS Congress. September 10–14, 2017. Jerusalem, Israel. *The FEBS Journal*. 2017; vol. 284 (S1), pp 144, P.1.5-073. DOI: 10.1111/febs.14174
- Göksoy E, **Demirkaya S**, Oflaz S, Özge G, Mumcuoğlu T, Can Demirdöğen B. Analysis of the relationship between PON1 (paraoxonase1) 192Q/R (rs662) polymorphism with pseudoexfoliation syndrome and pseudoexfoliative glaucoma. Erciyes Medical Genetics Days 2017. May 11–13, 2017. Kayseri, Turkey. *Erciyes Medical Journal*. 2017; vol 39, Issue 2, pp 13.
- **Demirkaya S**, Özge G, Mumcuoğlu T, Can Demirdöğen B. Evaluation of rs11136000 single nucleotide polymorphism of clusterin gene in pseudoexfoliation syndrome risk. The 41st FEBS Congress. September 3-8, 2016. *The FEBS Journal* 283 (Suppl. 1), 2016; pp 375, P-08.02.5-006. DOI: 10.1111/ febs.13808 (Abstract published in the *FEBS Journal* but the poster presentation could not be done because of the cancellation of the congress).

## **Skills**

- **Programming:** Python, JavaScript, R, HTML, MySQL, PostgreSQL, SQLite
- **Technologies & Libraries:** Docker, CWL, Git, Nextflow, 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)

## **Languages**

- Turkish (native)
- English (fluent)

## **Test Scores**

- ALES (Academic Personnel and Postgraduate Education Entrance Exam) – 82,8  
ÖSYM – 11.2017
- METU EPE (English Proficiency Exam) – 82,0  
Middle East Technical University – 06.2018
- YDS (Foreign Language Test) – 87,5 (Class B)  
ÖSYM – 04.2018

## **References**

Amit Jain – Graph Technology Director  
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