DR. ELMASNUR YILMAZ

PhD · RNA Biologist & Computational Genomicist · Neurogenetics
elmasnrylmz@gmail.com · +90 545 460 07 91 · ORCID: 0000-0001-9711-0203

PROFESSIONAL SUMMARY

PhD in Biotechnology with a rare, fully integrated profile spanning advanced wet-laboratory methods and computational genomics. Core expertise in TDP-43-mediated regulation of alternative splicing and cryptic exon inclusion, studied in the context of ALS/FTD neurodegeneration through end-to-end RNA-seq pipelines (rMATS, MAJIQ, DESeq2) and custom Python and R workflows. Extensive prior background in neurogenetic rare-disease diagnostics via whole-exome sequencing, yielding high-impact collaborative publications in Brain, iScience, and Neurology Genetics. Recipient of the highly competitive TUBITAK 2211-A National PhD Fellowship and the YOK 100/2000 Research Fellowship.

RESEARCH INTERESTS

RNA biology and post-transcriptional regulation · Alternative splicing and cryptic exon inclusion · TDP-43 and RNA-binding protein dysfunction in ALS/FTD · Alternative polyadenylation · Computational transcriptomics and reproducible pipeline development · Rare neurogenetic disease genomics and variant interpretation · Integration of wet-lab and dry-lab approaches.

EDUCATION

Ege University, Graduate School of Natural and Applied Sciences, Izmir, Turkiye Sep 2020 - Aug 2026
Ph.D. in Biotechnology
Dissertation: "Investigation of TDP-43 Mediated Regulation of Cryptic Exons and Identification of New Candidate Genes."
Advisor: Prof. Dr. Yasemin Eraç, Department of Pharmacology, Faculty of Pharmacy.

Dokuz Eylul University, Izmir Biomedicine and Genome Institute (IBG), Izmir, Turkiye Dec 2018
M.Sc. in Molecular Biology and Genetics (Neurogenetics Track)
Thesis: "New Genomic Approaches to Explore the Neurogenetic Disease Burden of Consanguineous Marriages in Turkey."
Advisor: Dr. Yavuz Oktay.

Istanbul Technical University (ITU), Istanbul, Turkiye Feb 2016
B.Sc. in Molecular Biology and Genetics
Thesis: "The Usage of Salt Leaching Method in the Construction of Porous Tissue Culture Scaffolds."
Advisor: Prof. Dr. Fatma Neşe Kök.

RESEARCH EXPERIENCE

Ege University, Eraç Lab, Faculty of Pharmacy, Izmir, Turkiye Sep 2020 - Aug 2026
PhD Researcher, Transcriptomics, RNA Biology & Pharmacological Cell Biology

- Designed and executed end-to-end bulk RNA-seq pipelines, from raw FASTQ retrieval (ENA/SRA) through quality control (FastQC, MultiQC), adapter trimming (Trim Galore), alignment (HISAT2, STAR), and post-processing (SAMtools, Picard), to detect TDP-43-dependent cryptic exon inclusion and differential splicing events transcriptome-wide using rMATS and MAJIQ.
- Identified and characterized alternative polyadenylation (APA) site dysregulation upon TDP-43 depletion, integrating bioinformatics outputs with custom R and Python visualization pipelines; presented findings at international TDP-43 disease conferences.

- Performed comprehensive differential expression analysis (DESeq2, edgeR), pathway enrichment (clusterProfiler, GO/KEGG), and publication-quality visualization (ggplot2, ComplexHeatmap, EnhancedVolcano, seaborn); generated reproducible reports in R Markdown and Jupyter Notebooks.
- Maintained wet-laboratory experiments in parallel: routine cell culture (HEK293, cancer cell lines), MTT/WST-1 cytotoxicity assays, calcium imaging, pharmacological drug-response experiments, and phytochemical extract-based anticancer mechanism studies.
- Built reproducible bioinformatics workflows with Snakemake and Nextflow on Unix/Linux HPC clusters (SLURM); managed conda/mamba environments and documented code on GitHub for reproducibility.

Newcastle University, Institute of Genetic Medicine / International Centre for Life, UK Jun - Jul 2018

Visiting Researcher, Rare Neurogenetic Disease Diagnostics (Host: Prof. Rita Horvath)

- Analyzed whole-exome sequencing (WES) data from pediatric patients with rare neurological diseases; contributed to variant prioritization and interpretation pipelines used in the international CONSEQUITUR consortium.
- Cultured patient-derived fibroblasts and neural progenitor cells (NPCs); performed western blotting to assess protein expression in disease-relevant models.

Dokuz Eylul University, Oktay Lab, Izmir Biomedicine and Genome Center (IBG) Feb 2016 - Dec 2018

MSc Researcher, Neurogenetic Rare-Disease Genomics & Functional Validation

- Performed WES analysis on consanguineous Turkish families with severe pediatric neurogenetic conditions (mitochondrial disease, cortical malformations, neurodevelopmental disorders); identified pathogenic variants validated by Sanger sequencing and published in high-impact journals including Brain.
- Applied runs-of-homozygosity (RoH) analysis to map autozygous regions and prioritize recessive candidate variants; generated and interpreted VCF, BED, and GTF files for downstream analysis.
- Executed CRISPR-Cas9 experiments for in-cell validation of candidate pathogenic variants; performed DNA/RNA extraction, PCR, gel electrophoresis, and Sanger sequencing from patient blood samples.
- Contributed to the RD-Connect platform-based global rare-disease network, presenting findings at ICNC 2018 (Mumbai, India) and MolBiyoKon 2018 (Izmir, Turkey).

Istanbul Technical University, Kök Lab Feb - Jun 2015

Undergraduate Researcher, Tissue Engineering

- Fabricated porous scaffolds via salt-leaching for tissue culture applications; characterized structural properties for cell attachment and proliferation experiments.

TUBITAK, Marmara Research Center (MAM), Genetic Engineering and Biotechnology Institute Jun - Jul 2014

Research Intern

- Monoclonal antibody production via hybridoma technology; cell culture (passage, freezing, counting) and indirect ELISA assays.

Marmara University, Department of Bioengineering, Istanbul, Turkiye Aug - Sep 2013

Intern, Bioengineering

- Project: “Investigation of Beta-Lactamase Allosteric Site Ligand Recognition and Binding”; performed primer design, SDS-PAGE, sonication, and enzyme activity assays.

Royal Institute of Technology (KTH), Stockholm, Sweden Jun 2011

Summer Program Participant, “S(t)imulating Life: From CPU to Your Stomach”

- Introduction to molecular dynamics simulation using GROMACS; protein structure and simulation analysis, an early exposure to computational biology.

PEER-REVIEWED PUBLICATIONS

Author name underlined denotes Elmasnur Yilmaz. Listed in reverse chronological order.

8. Yasar, B.Y., Yilmaz, E., & Erac, Y. (2026). Pharmacological Inhibition of Store-Operated Calcium Entry Enhances Sorafenib-Induced Antitumor Effects in Huh7 Hepatocellular Carcinoma Cells. *Cancer Chemotherapy and Pharmacology*, 96(1), Article 87. doi:10.1007/s00280-026-04934-5.

7. Demir, S., Yasar, B.Y., Yilmaz, E., Karasu, A.Z., Erac, Y., & Baykan, S. (2025). Investigation of Cytotoxic Activity of *Salvia tomentosa* and Methyl Carnosate-12-Methylether on Different Cancer Cell Lines. *Revista Brasileira de Farmacognosia.*

6. Kurul, S.H., Oktay, Y., Topf, A., … Yilmaz, E., … & Horvath, R. (2021). High diagnostic rate of trio exome sequencing in consanguineous families with neurogenetic diseases. *Brain*.

5. Gungor, S., Oktay, Y., Hiz, S., … Yilmaz, E., … & Horvath, R. (2021). Autosomal recessive variants in TUBGCP2 alter the γ-tubulin ring complex leading to neurodevelopmental disease. *iScience*, 24(1), 101948.

4. Yaramis, A., Lochmüller, H., Töpf, A., Sonmezler, E., Yilmaz, E., Hiz, S., … & Beltran, S. (2020). COL4A1-related autosomal recessive encephalopathy in 2 Turkish children. *Neurology Genetics*, 6(1).

3. Oktay, Y., Güngor, S., Zeltner, L., … Yilmaz, E., … & Kaemereit, S. (2020). Confirmation of TACO1 as a Leigh Syndrome Disease Gene in Two Additional Families. *Journal of Neuromuscular Diseases*, 7(3), 301-308.

2. Töpf, A., Oktay, Y., Balaraju, S., Yilmaz, E., & Hiz, S., Horvath, R. (2019). Severe neurodevelopmental disease caused by a homozygous TLK2 variant. *European Journal of Human Genetics*. doi:10.17863/CAM.40761.

1. Yilmaz, E., Sonmezler, E., Töpf, A., Balaraju, S., Yaramis, A., Gungor, S., … & Hiz, S. (2019). A novel mutation in the coiled-coil interaction domain of LAMB1 extends the molecular basis of laminin-related cortical malformation phenotypes. *European Journal of Human Genetics*, 27(Suppl. 1), 285-286.

SUBMITTED MANUSCRIPTS

Yasar, B.Y., Yilmaz, E., Onal, F.N., Baykan, S., & Erac, Y. Antiproliferative Activity of *Morus nigra* L. Fruit Fractions in Oral Squamous Cell Carcinoma Cells: In Vitro Activity and Transcriptomic Context. Submitted to *Istanbul Journal of Pharmacy*.

Yasar, B.Y., Yilmaz, E., & Erac, Y. Integrative Bioinformatic Analysis of TRPC6 and STIM1 in Hepatocellular Carcinoma: Expression Patterns, Prognostic Associations, and Functional Dependencies. Submitted to *Eskisehir Technical University Journal of Science and Technology C - Life Sciences and Biotechnology*.

Okeer, M., Sezgin, S., Onal Hasar, F.N., Yilmaz, E., Aydemir, S.S., Baykan, S., & Erac, B. Effects of *Morus nigra* L., *Salvia tomentosa* Mill., and *Salvia dichroantha* Stapf. extracts on the virulence factors of *Stenotrophomonas maltophilia* and *Acinetobacter baumannii* isolates. Submitted to *Microbial Pathogenesis*. Manuscript No: YMPAT-D-26-02556.

CONFERENCE PRESENTATIONS

Oral Presentations

- Yilmaz, E., & Erac, Y. (2026). “Comparative Splicing Analysis of Calcium Signaling Genes upon TDP-43 Gene Silencing” [TDP-43 Gen Susturmasında Kalsiyum Sinyalizasyon Genlerinin Karşılaştırmalı Kırpılma Analizi]. 9th International Health Sciences and Life Congress (IHSLC 2026), Antalya, Türkiye, April 15–18, 2026. Oral presentation; full-text proceeding published in the congress proceedings book.
- Yilmaz, E. (2023). “Disruption of TDP-43 Expression Leads to Transcriptome-Wide Deregulation of Polyadenylation Sites.” ICGEB Arturo Falaschi Conference, 1st Biennial Conference on TDP-43 Function and Dysfunction in Disease, Trieste, Italy.
- Yilmaz, E. (2018). “Better Call RoH: What an 11 Mb Homozygous Haplotype Block Can Tell Us About a Possible Founder Mutation in the Turkish Population.” 6th International Congress of the Molecular Biology Association of Turkey (MolBiyoKon), Izmir, Turkey.
- Yilmaz, E. (2018). Molecular Genetics Contributions to Pediatric Neurology: Sequence Analysis via RD-Connect Platform. Neurology Seminar, Izmir, Turkey.

Poster Presentations

- Yilmaz, E., Erac, Y. (2024). "TDP-43 Mediated Splicing and Its Intersection with Ubiquitination in Gene Regulation." 3rd Ubiquitin Function in Health & Disease Conference, Lisbon, Portugal.
- Yilmaz, E., Yasar, B.Y., Getboga, D., Sen, T.D., Karstarli, I.N., Erac, Y. (2024). "The Effects of Hyperforin in Sorafenib-Resistant Hepatocellular Carcinoma." EPHAR 2024, 9th European Congress of Pharmacology, Athens, Greece.
- Yilmaz, E., Peng, L., Moore, M.J., Kaymaz, Y. (2023). "Disruption of TDP-43 Expression Leads to Transcriptome-Wide Deregulation of Polyadenylation Sites." ICGEB Arturo Falaschi Conference on TDP-43, Trieste, Italy. *[Also selected for oral presentation]*
- Hiz, S., Yilmaz, E., Oktay, Y. (2018). CONSEQUITUR: A Multinational Effort Towards Determining the Neurogenetic Disease Burden of Consanguineous Marriages in Turkey. 15th International Child Neurology Congress (ICNC), Mumbai, India.
- Gungor, S., Okur, D., Yilmaz, E., Hiz, S. (2018). Two Distinct Phenotypes Caused by the Same Mutation in the SAMHD1 Gene. ICNC 2018, Mumbai, India.
- Töpf, A., Oktay, Y., Balaraju, S., Yilmaz, E., & Horvath, R. (2018). Unexpected Genetic Diagnosis of Mitochondrial Disease in Three Consanguineous Turkish Families. *Neuromuscular Disorders*, 28, S85-S86.

FELLOWSHIPS, SCHOLARSHIPS & AWARDS

TUBITAK 2211-A National PhD Scholarship (Highly Competitive) Sep 2021 - Aug 2025
Scientific and Technological Research Council of Turkiye, awarded to top-ranked PhD candidates nationally.

YOK 100/2000 Molecular Pharmacology & Pharmaceutical Research Fellowship Sep 2020 - Aug 2024
Council of Higher Education of Turkiye, competitive national research fellowship program.

TUBITAK Project Scholarship, Neurogenetic Disease Burden Study (Project #216S771) Oct 2017 - Dec 2018
Funded MSc research on WES-based diagnosis of rare neurogenetic diseases in consanguineous families.

TUBITAK Project Scholarship, Glioma SNP Mechanisms Study (Project #214S097) Nov 2016 - Sep 2017
Funded research on molecular mechanisms of SNP rs55705857 predisposing to gliomas in 8q24.21.

RESEARCH PROJECTS

splicescope — Open-source alternative-splicing analysis toolkit 2026 · v0.5.0
Python toolkit turning splice junctions into a junction taxonomy, splice-site usage (Ψ), event-level PSI for SE/MXE/A5SS/A3SS, differential splicing (ΔΨ + FDR), pathway over-representation, and ML-based cryptic-event calling; unit tests, GitHub Actions CI, a Nextflow pipeline and a Streamlit app. github.com/elmasnuryilmaz/splicescope

Researcher, TUBITAK Project #124S480 Oct 2024 - Oct 2025
"Effects of Morus nigra L., Salvia tomentosa Mill. and Salvia dichroantha Stapf. Extracts on Virulence Factors of Stenotrophomonas maltophilia and Acinetobacter baumannii."

Researcher, Ege University BAP Project #31901 Sep 2023 - Sep 2025
"Investigation of TDP-43 Mediated Regulation of Cryptic Exons and Identification of New Candidate Genes."

Researcher, Ege University BAP Project #29657 May 2023 - May 2024
"Investigation of Anticancer Effect and Mechanism of Salvia tomentosa Extracts on Different Cancer Cell Lines."

Researcher, TUBITAK Project #216S771 Oct 2017 - Dec 2018
"New Genomic Approaches to Explore the Neurogenetic Disease Burden of Consanguineous Marriages in Turkey."

Researcher, TUBITAK Project #214S097 Nov 2016 - Sep 2017
"Uncovering the Molecular Mechanisms of the SNP rs55705857 in the 8q24.21 Region Predisposing to Gliomas."

TECHNICAL SKILLS

Bioinformatics & Computational Biology

RNA-seq Analysis: FastQC, MultiQC, Trim Galore, Cutadapt, HISAT2, STAR, SAMtools, BAMtools, Picard

Alternative Splicing & Cryptic Exons: rMATS, MAJIQ; splice junction visualization and exon usage quantification

Transcript Quantification: featureCounts, HTSeq-count, StringTie; GENCODE/Ensembl GTF/GFF annotation

Differential Expression: DESeq2, edgeR, limma; PCA, hierarchical clustering, volcano plots

Pathway & Functional Enrichment: clusterProfiler, GO, KEGG, EnhancedVolcano, ComplexHeatmap

Alternative Polyadenylation (APA): APA site analysis and transcriptome-wide polyadenylation deregulation detection

Variant Analysis & Rare-Disease WES: VCF interpretation, runs-of-homozygosity (RoH), variant prioritization, Sanger validation

Genome Coordinate Tools: BEDTools, GTFtools, GffCompare; BED, GTF, VCF, BAM formats

Data Retrieval: ENA/SRA (prefetch, fasterq-dump); FASTQ/BAM organization and indexing

Programming & Scripting

Python: Data wrangling and visualization (Pandas, NumPy, Seaborn, Matplotlib, plotnine); Jupyter Notebooks (VSCode)

R: Statistical genomics (DESeq2, ggplot2, tidyverse, ComplexHeatmap, RStudio); R Markdown for reproducible reports

Shell / Bash: Workflow automation, batch processing, HPC job scripting on Unix/Linux systems

Workflow Management & Computing

Pipeline Development: Snakemake, Nextflow; reproducible and scalable bioinformatics pipelines

HPC / Cloud: Institutional HPC clusters (SLURM job scheduling), Google Colab (GPU-accelerated notebooks)

Environment Management: Conda, Mamba; troubleshooting tool installations on Mac M1 / Apple Silicon

Version Control: Git, GitHub; version control, collaboration, reproducible research documentation

Wet Laboratory Skills

Cell Biology: Mammalian cell culture (HEK293, cancer lines, fibroblasts, neural progenitor cells); passage, freezing, transfection

Molecular Biology: PCR, qPCR, gel electrophoresis, western blotting, Sanger sequencing, DNA/RNA extraction from blood

Functional Assays: MTT/WST-1 viability, calcium imaging, pharmacological drug-response assays, indirect ELISA

Gene Editing: CRISPR-Cas9 design and execution for in-cell variant validation

Genomic Methods: Whole-exome sequencing (library prep familiarity); blood collection and biobanking protocols

CERTIFICATIONS & PROFESSIONAL DEVELOPMENT

Computational Genomics Course 2025 (Compgen2025), Module 1: AI-Assisted Data Analysis — Mar 2025

Compgen2025, Module 3: Multi-omics Data Integration & Deep Learning in Precision Oncology — Mar 2025

Fundamentals of AI/ML in Precision Medicine, Stanford Deep Data Research Center — Apr 2024

Fundamentals of Data Science in Precision Medicine and Cloud Computing, Stanford DDRC — Apr 2024

LANGUAGE PROFICIENCY

English: C1 Advanced. YOKDIL: 96.25/100 (2022); IELTS: 7.0 Overall (Listening 7.0, Reading 8.0, Writing 6.0, Speaking 6.0; 2018).

Turkish: Native.

PROFESSIONAL MEMBERSHIPS

- European Association for Cancer Research (EACR), Member #EACR35041.

- Alzheimer's Association International Society to Advance Alzheimer's Research and Treatment (ISTAART), Member #100020374703.
- Istanbul Technical University Alumni Association, Active Mentor.