

# ELMASNUR YILMAZ

## PHD CANDIDATE

Mail: elmasnrylmz@gmail.com  
ORCID: 0000-0001-9711-0203

İzmir, Türkiye

## EDUCATION

---

### *Ph.D.*

#### **Ege University**

Graduate School of Natural and Applied Sciences

Department of Biotechnology

September 2020 – cont.

Thesis Proposal Title: “Investigation of TDP-43 Mediated Regulation of Cryptic Exons and Identification of New Candidate Genes”

Advisor: Prof. Dr. Yasemin ERAÇ

### *Master's Degree*

#### **Dokuz Eylul University**

Izmir Biomedicine and Genome Institute

Molecular Biology and Genetics

December 2018

Thesis: “New genomic approaches to explore the neurogenetic disease burden of consanguineous marriages in Turkey”

Advisor: Dr. Yavuz OKTAY

### *Bachelor's Degree*

#### **Istanbul Technical University**

Molecular Biology and Genetics

February 2016

Thesis: “The usage of salt leaching method in the construction of porous tissue culture scaffolds”

Advisor: Prof. Dr. Fatma Neşe KÖK

## SCHOLARSHIPS

---

### **BİDEB (The Scientific and Technological Research Council of Türkiye) 2211-A National PhD Scholarship Program**

September 2021– August 2025

### **YÖK (Council of Higher Education of Türkiye) 100/2000 Molecular Pharmacology and Pharmaceutical Research**

Fellow

September 2020 – August 2024

### **TUBITAK Project Scholar**

"New Genomic Approaches to Explore the Neurogenetic Disease Burden of Consanguineous Marriages in Turkey"

### **TUBITAK Project Scholar**

"Uncovering the Molecular Mechanisms of the SNP rs55705857 in the 8q24.21 Region Predisposing to Gliomas"

## PROJECTS

---

### **Researcher Ege University BAP**

**Project Number: 31901**

“Investigation of TDP-43 Mediated Regulation of Cryptic Exons and Identification of New Candidate Genes”

September 2023-September 2025

### **Researcher Ege University BAP**

**Project Number: 29657**

“Investigation of Anticancer Effect and Mechanism of Extracts from *Salvia tomentosa* Above Ground Parts on Different Cancer Cell Lines”

May 2023- May 2024

**Researcher TUBITAK**  
**Project Number: 216S771**

October 2017 – December 2018

"New Genomic Approaches to Explore the Neurogenetic Disease Burden of Consanguineous Marriages in Turkey"

**Researcher TUBITAK**  
**Project Number: 214S097**

November 2016 – September 2017

"Uncovering the Molecular Mechanisms of the SNP rs55705857 in the 8q24.21 Region Predisposing to Gliomas"

## EXPERIENCE

---

**Newcastle University**, Newcastle upon Tyne, United Kingdom Jun 2018/Jul 2018  
**Visiting Student**, Institute of Genetic Medicine/International Centre for Life  
Whole exome sequencing data analysis, fibroblast cell culture, western blotting, neural progenitor cell studies

**Dokuz Eylul University**, İzmir, Türkiye Feb 2016/Dec 2018  
**Researcher**, Oktay Lab, İzmir Biomedicine and Genome Center  
Collection of blood samples from patients, isolation of DNA and RNA from samples, preliminary screening with routine PCR and gel electrophoresis, Sanger sequence analysis, whole exome sequencing data analysis, routine cell culture studies and analysis, CRISPR-Cas studies

**Istanbul Technical University**, İstanbul, Türkiye Şub 2015/Haz 2015  
**Undergraduate Researcher**, Kök Lab  
"The usage of salt leaching method in the construction of porous tissue culture scaffolds"  
Basic molecular methods in tissue engineering laboratory for undergraduate thesis

**TUBİTAK/MAM**, Kocaeli, Türkiye Jun 2014/Jul 2014  
**Intern**, Genetic Engineering and Biotechnology Institute  
"Production of Monoclonal Antibody Using Hybridoma Technology"  
Cell culture studies (passage, counting, freezing), indirect ELISA

**Marmara University**, İstanbul, Türkiye Aug 2013/Sep 2013  
**Intern**, Bioengineering  
"Investigation of Beta-Lactamase Allosteric Site Ligand Recognition and Binding"  
Primer design, SDS-PAGE, sonication, and enzyme activity assays

**Royal Institute of Technology (KTH)**, Stockholm, Sweden Jun 2011  
**Participant**  
"S(t)imulating life: from CPU to your stomach"  
Protein simulation using GROMACS

## PUBLICATIONS

---

Kurul, S. H., Oktay, Y., Töpf, A., Szabó, N. Z., Güngör, S., Yaramis, A., ... **Yilmaz, E.**, ... & Horvath, R. (2021). High diagnostic rate of trio exome sequencing in consanguineous families with neurogenetic diseases. *Brain*.

Gungor, S., Oktay, Y., Hiz, S., Aranguren-Ibáñez, Á., Kalafatcilar, I., Yaramis, A., ... **Yilmaz, E.**, ... & Horvath, R. (2021). Autosomal recessive variants in TUBGCP2 alter the  $\gamma$ -tubulin ring complex leading to neurodevelopmental disease. *Isience*, 24(1), 101948.

Oktay, Y., Gungor, S., Hiz, S., Yaramis, A., Aranguren-Ibanez, A., Yis, U., ... **Yilmaz, E.**, ... & Horvath, R. (2020). A homozygous missense variant in TUBGCP2 alter the  $\gamma$ -tubulin ring complex leading to abnormal cortical development, pontocerebellar atrophy and altered myelination. *European Journal Of Human Genetics*, 28(Suppl 1), 460–461.

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).

Oktay, Y., Güngör, S., Zeltner, L., Wiethoff, S., Schöls, 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.

Topf, 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*. 2019 June. doi:10.17863/CAM.40761.

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

## POSTERS

---

**Elmasnur Yilmaz**, Yasemin Erac (2024, July). “Tdp-43 Mediated Splicing and Its Intersection with Ubiquitination In Gene Regulation” 3rd Ubiquitin Function in Health & Disease Conference 07 – 10 July 2024 Lisboa, Portugal.

**Elmasnur Yilmaz**, Beste Yurdacan Yasar, Damla Getboga, Turkum Dilan Sen, Ilay Nur Karstarli, Yasemin Erac (2024, June). “The Effects of Hyperforin in the Sorafenib-Resistance Hepatocellular Carcinoma” EPHAR 2024 The Federation of European Pharmacological Societies 9<sup>th</sup> European Congress of Pharmacology 23-26 June 2024 Athens, Greece.

**Elmasnur Yilmaz**, Lingtao Peng, Melissa J Moore, Yasin Kaymaz (2023, Sep). “Disruption of TDP43 expression leads to transcriptome-wide deregulation of polyadenylation sites” ICGEB Arturo Falaschi Conference "1st biennial conference on TDP-43 function and dysfunction in disease" 06/09/2023 to 08/09/2023 Trieste, Italy.

Hiz, S., **Yilmaz, E.**, & Oktay, Y. (2018, Nov). CONSEQUITUR: A multinational effort towards determining the neurogenetic disease burden of consanguineous marriages in Turkey by genomics approaches. Poster presented at the 15th International Child Neurology Congress (ICNC 2018), Mumbai, India.

Gungor, S., Okur, D., **Yilmaz, E.**, & Hiz, S. (2018, Nov). Two distinct phenotypes caused by the same mutation in the SAMHD1 gene. Poster presented at the 15th International Child Neurology Congress (ICNC 2018), Mumbai, India.

Topf, A., Oktay, Y., Balaraju, S., **Yilmaz, E.**, & Horvath, R. (2018). MITOCHONDRIAL DISEASES (Posters): P.193 Unexpected genetic diagnosis of mitochondrial disease in three consanguineous Turkish families. *Neuromuscular Disorders*, 28, s85-s86.

## TALKS

---

**Elmasnur YILMAZ** (2023, Sep). “Disruption of TDP43 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, 06/09/2023 to 08/09/2023.

**Elmasnur YILMAZ** (2018, Sep). “Better Call RoH: What a 11Mb 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 Conference 2018), Izmir, Turkey.

**Elmasnur YILMAZ** (2018, Dec). Moleküler Genetikteki Son Gelişmelerin Çocuk Nöroloji Hastalıklarının Tanı ve Tedavisine Katkıları: RD-Connect Platform Aracılığıyla Sekans Analizi. (Contributions of Latest Developments in Molecular Genetics to the Diagnosis and Treatment of Child Neurology Diseases: Sequence Analysis via RD-Connect Platform). Neurology Seminar Izmir, Turkey

## LANGUAGE

---

**English:** CEFR Level C1 IELTS Test Result:7 (Listening: 7.0 Reading: 8.0 Writing: 6.0 Speaking: 6.0 Test Date: January 2018)

YÖKDİL English Exam Result: 96.25/100 (Exam Date: 13.03.2022)

## PROFESSIONAL MEMBERSHIPS

---

- European Association for Cancer Research (EACR) Membership Number: EACR35041
- The Alzheimer's Association International Society to Advance Alzheimer's Research and Treatment (ISTAART) Membership Number: 100020374703
- Istanbul Technical University Alumni Association (Mentor)

## SKILLS

---

- Proficiency in programming languages commonly used in bioinformatics such as Python, R, and Java.
- Experience with bioinformatics tools and databases such as BLAST, FASTQC, SAMtools.
- Proficiency in using Linux/Unix and command line interface tools.
- Familiarity with molecular modeling and simulation tools.
- Expertise in next-generation sequencing data analysis.
- Knowledge of bioinformatics pipelines and workflows.
- Experience with data visualization and interpretation to communicate results to non-technical audiences.
- Knowledge of molecular biology and genetics concepts and techniques.
- Effective collaboration and communication skills for working with interdisciplinary teams.
- Experimental design: Ability to design and execute experiments using various techniques such as molecular biology, cell biology, biochemistry, and microscopy.
- Laboratory techniques: Proficiency in a variety of laboratory techniques such as DNA/RNA isolation, PCR, gel electrophoresis, cloning, cell culture.
- Laboratory management: Experience in managing laboratory operations, including ordering supplies, maintaining equipment, and ensuring compliance with safety regulations.
- Critical thinking and problem solving: Ability to identify and solve complex problems in experimental design, data interpretation, and troubleshooting.
- Project management: Experience in managing research projects from start to finish, including project planning, budgeting, and execution.
- Scientific writing and communication: Proficiency in writing scientific papers, reports, and presentations for peer-reviewed journals and conferences.
- Mentoring and training: Experience in mentoring students and junior researchers, and training lab personnel on experimental protocols.