CURRICULUM VITAE

Name: Kaya Bilguvar, M.D., Ph.D.

Education:

M.D.	Marmara University School of Medicine, Istanbul, Turkey, 2000
Ph.D.	Marmara University Health Sciences Institute, Istanbul, Turkey, 2022

Career/Academic Appointments:

2001-2002	Research Intern, Department of Medical Biology and Genetics, Marmara University Health Sciences Institute, Istanbul, Turkey
2005-2007	Post-doctoral Research Associate, Department of Neurosurgery, Yale School of Medicine, New Haven, CT
2007-2013	Associate Research Scientist, Department of Neurosurgery, Program on Neurogenetics, Yale School of Medicine, New Haven, CT
2013-2019	Assistant Professor, Department of Genetics, Yale School of Medicine, New Haven, CT
2019-2021	Associate Professor with term, Department of Genetics, Yale School of Medicine, New Haven, CT
2020-2021	Associate Professor, Department of Neurosurgery, Yale School of Medicine, New Haven, CT
2021-2023	Specialist, Department of Medical Genetics, School of Medicine, Acibadem University, Istanbul, Turkey
2021-present	Associate Professor Adjunct, Department of Genetics, Yale School of Medicine, New Haven, CT
2021-present	Associate Professor Adjunct, Department of Neurosurgery, Yale School of Medicine, New Haven, CT
2023-present	Assistant Professor, Department of Medical Genetics, School of Medicine, Acibadem University, Istanbul, Turkey

Administrative Positions:

2013-2016	Associate Director, Yale Center for Genome Analysis, Yale School of Medicine, New Haven, CT
2016-2021	Director, Yale Center for Genome Analysis, Yale School of Medicine, New Haven, CT
2023-present	Chair, Department of Medical Genetics, School of Medicine, Acibadem University, Istanbul, Turkey
2023-present	Chair, Translational Medicine Program, Institute of Health Sciences, Acibadem University, Istanbul, Turkey
2024-present	Associate Director, Institute of Health Sciences, Acibadem University, Istanbul, Turkey

Professional Honors & Recognition:

International/National:

2010 Runner-up, Scientific Breakthrough of the Year, the journal *Science 17 Dec 2010: Vol. 330, Issue 6011, pp. 1605-1607* (one of the first development and application of whole-exome sequencing to human disease; Nature. 2010 Sep 9;467(7312):207-10)

Active Grants:

Agency: NIH/NICHD USA
I.D.# R01 HD110693
Title: A Gene-Network Discovery Approach to Structural Brain Disorders
P.I.s: Kaya Bilguvar, Angeliki Louvi
Role on Project: Principal investigator
Percent effort: 25%
Total costs for project period: $3,328,715
Project period: 09/15/2023 – 05/31/2028

Agency: Turkish Scientific and Technological Research Council (TUBITAK)
I.D.# ARDEB 1001-222S389
Title: Investigation and Molecular Characterization of Obesity Related Genes STEAP1B, KCNQ5 and UCP1
P.I.s: Onur Emre Onat
Role on Project: Co-investigator
Percent effort: 10%
Total costs for project period: 2,048,806TL
Project period: 07/15/2023 – 07/15/2026

Agency: Turkish Scientific and Technological Research Council (TUBITAK)
I.D.# ARDEB 1001-221S889
Title: Deep-CP: Development of a Deep Learning Variant Pathogenicity Prediction Tool using AlphaFold for Cerebral Palsy
P.I.s: Gunseli Bayram Akcapinar
Role on Project: Co-investigator
Percent effort: 25%
Total costs for project period: 771,750TL
Project period: 04/15/2022 – 04/15/2025

Agency: European Commission Horizon Europe Framework Programme Call: HORIZON-WIDERA-2021-ACCESS-03-01 (Twinning)
I.D.# 101078981

Title: GEMSTONE -Genetically Engineering Experimental Models: Enhancement of scientific and technological excellence and innovation potential to study neurodevelopmental diseases
P.I.s: Filiz Onat
Role on Project: Co-investigator (Task. 2.2)
Percent effort: %5
Total costs for project period: €1,450,446.45
Project period: 01/10/2021 – 30/09/2025

Past Grants:

Agency: NIH/NINDS USA
I.D.# R01 NS111029
Title: Human genetics and molecular mechanisms of congenital hydrocephalus
P.I.s: Kristopher Kahle
Role on Project: Co-investigator
Percent effort: 5%
Total costs for project period: $2,460,261
Project period: 02/01/2020 – 01/31/2025

Agency: Istanbul Development Agency (ISTKA)
I.D.# TR10/22/TNH
Title: Istanbul Rare and Undiagnosed Disease Platform
P.I.s: Ugur Ozbek
Role on Project: Co-investigator
Percent effort: 10%
Total costs for project period: 12,987,013.03TL
Project period: 09/19/2022 – 09/19/2024

Agency: NIH/NINDS USA
I.D.# R01 NS106298
Title: Genomic insights into the neurobiology of cerebral palsy
P.I.s: Kaya Bilguvar, Michael Kruer (University of Arizona Health Sciences)
Role on Project: Principal investigator for Yale
Percent effort: 20%
Total costs for project period: $372,293 (allocated to Yale)
Project period: 02/01/2019 – 01/31/2024

Agency: NIH/NIDDK USA
I.D.# P30 DK079310
Title: George M. O'Brien Kidney Center at Yale
P.I.s: Peter Aronson
Role on Project: Co-investigator
Percent effort: 12%
Total costs for project period: $713,865
Project period: 09/20/2018 – 07/31/2023

Agency: NIH/NHGRI/NHLBI USA
I.D.# UM1 HG006504
Title: "Yale Center for Mendelian Genomics"
P.I.s: Richard Lifton, Shrikant Mane, Murat Gunel, Mark Gerstein
Role on Project: Co-investigator
Percent effort: 15%
Total costs for project period: $11,902,168
Project period: 12/01/2015 – 11/30/2021

Agency: NIH/NIMH USA
I.D.# R01 MH102342
Title: Integrating the genomics of Autism Spectrum Disorders (ASD) in consanguineous and "idiopathic" families
P.I.s: Murat Gunel, Matthew State
Role on Project: Co-investigator
Percent effort: 10%
Total costs for project period: $2,427,874
Project period: 05/08/2015 – 02/28/2019

Agency: NIH/NIMH USA
I.D.# R01 MH103616
Title: Disease Gene Discovery in Structural Brain Disorders
P.I.s: Kaya Bilguvar, Murat Gunel
Role on Project: Principal investigator
Percent effort: 10%
Total costs for project period: $1,485,239
Project period: 04/05/2014 – 03/31/2017

Agency: NIH/NINDS/NHLBI USA
I.D.# R01 NS057756
Title: Molecular Genetic Pathogenesis of Intracranial Aneurysms
P.I.s: Murat Gunel
Role on Project: Co-investigator
Percent effort: 10-50%
Total costs for project period: $5,280,380
Project period: 03/01/2007 – 02/28/2017

Agency: NIH/NHGRI/NHLBI USA
I.D.# U54 HG006504
Title: Yale Center for Mendelian Genetics
P.I.s: Richard Lifton, Shrikant Mane, Murat Gunel, Mark Gerstein
Role on Project: Co-investigator
Percent effort: 46.66%
Total costs for project period: $10,800,000
Project period: 12/05/2011 – 11/30/2015

Agency: NIH/NINDS
I.D.# R01 NS067023
Title: Molecular Variants that Determine Genetic Susceptibility to Intracranial Aneurysm
P.I.s: Murat Gunel
Role on Project: Co-investigator
Percent effort: 25-50%
Total costs for project period: $3,148,628
Project period: 09/30/2009 – 07/31/2015

Agency: NIH/NINDS/NIMH USA
I.D.# RC2 NS070477
Title: Gene Discovery in Recessive Structural Brain Disorders through Whole Exome Sequencing
P.I.s: Murat Gunel
Role on Project: Co-investigator
Percent effort: 25-50%
Total costs for project period: $2,906,138
Project period: 09/30/2009 – 02/29/2012

Invited Speaking Engagements, Presentations, Symposia & Workshops Not Affiliated with Yale and Acibadem Universities:

International/National:

2025	9th International Cerebral Palsy and Developmental Disorders Congress, Istanbul, Turkey: "Genetics of Neurodevelopmental Disorders"
2025	Turkish Association of Histology and Embryology From Outside the Borders Within the Labs Seminar Series, Istanbul, Turkey: "Genetics and Modeling of Neurodevelopmental Disorders: What Have I Been Doing for the Last 30 Years?"
2025	Molecular Biology and Genetics Turkey From Local to Global Webinar Series, Istanbul, Turkey: "From Genetics to Modeling: My 20plus-year Journey into the Nervous System"
2024	Marmara University School of Medicine Pediatrics Seminars, Istanbul, Turkey: "Genetics and Modeling: "Neurodevelopmental Disorders from Mendelian to Complex"
2024	22nd National Neuroscience Congress, Istanbul, Turkey: "Multi-omic Approaches and Gene Discovery In Neurological Disorders"
2023	Hacettepe University Neurological Sciences and Psychiatry Institute Seminar Series, Ankara, Turkey: "Genetics, Modeling and Beyond: 20 years on neurodevelopmental disorders"
2023	9th International BAU Drug Design Congress, Istanbul, Turkey: "Common Diseases, Rare Variants, Unique Patients: towards individualized treatments"
2023	Ege University Medical School 5th Research Education Festival, Izmir, Turkey: "Genetics, Modeling and Beyond: 20 years on neurodevelopmental disorders"

2023 59th National Neurology Congress, Antalya, Turkey: "Personalized Disease Modeling: Organoids"
2023 Uskudar University Scientific World Summit on Green Transformation, Animal Rights and Veganism, Istanbul, Turkey: "Genetics and Modeling of Neurodevelopmental Disorders"
2023 Bezmialem University BILSAB Seminar Series, Istanbul, Turkey: "Genetics, Modeling and Beyond: 20 years on neurodevelopmental disorders"
2023 8th Erciyes Medicine Medical Genetics Congress, Kayseri, Turkey: "Appropriate Evaluation of Genetic Results by the Clinician"
2023 7th Neurogenetics Course of Turkish Association of Neurology, Istanbul, Turkey: "Next-generation sequencing and Modeling Approaches to Neurodevelopmental Disorders"
2023 10th Marmara Pediatric Congress, İstanbul, Turkey: "Genetics and Modeling of Neurodevelopmental Disorders"
2022 20th National Neuroscience Congress, Istanbul, Turkey: "Genetics and Modeling of Pediatric Brain Disease"
2021 International Conference of the Genetics Society of Korea, Seoul, Korea: "Genetics and Modeling of Pediatric Brain Disease"
2018 Bioinformatics Forum, Gebze, Turkey: "Genome Profiling for The Clinics: Options, Timing, Expectations, Hurdles"
2017 Turkish Medical World Congress, Istanbul, Turkey: "Research-Clinical Interface for Genomics of Newborn Disorders"
2017 Turkish Medical World Congress, Istanbul, Turkey: "Cancer Genetics: Yale Center for Genome Analysis Experience"
2017 22nd National Cancer Congress, Antalya, Turkey: "Genomic Testing in Cancer: Choices and Applications"
2016 3rd Turkish Medical World Congress, Istanbul, Turkey: "Personalized Medicine Applications and the National Genome Project"
2016 Neurometabolic Dysmorphology Symposium, Istanbul, Turkey: "New generation genomic technologies and applications in health"
2015 Symposium of TURKMSIC, Istanbul, Turkey: "Genomic Sequencing Technologies and Clinical Applications"
2014 MipTec, Basel, Switzerland: "Rapid sequencing and clinical application of pediatric brain tumor genomes"
2013 International Symposium of the Association of Biomolecular Resource Facilities, Palm Springs, CA: "Whole-exome Sequencing in Nervous System Disorders"

Peer-Reviewed Presentations & Symposia Given at Meetings Not Affiliated with Yale and Acibadem Universities:

International/National:

2025 Europe Biobank Week 2025, Bologna, Italy: "Integrating Standardized Biobanking into Rare Disease Research: Insights from the ISTisNA Platform"

2024	The European Workshop For A Multidisciplinary View On Rare Genetic Neurodevelopmental Disorders (EuroNDD), Lisbon, Portugal: "Identifying Novel Candidate Genes and Variants in a Turkish Cerebral Palsy Cohort: Utilizing Whole and Clinical Exome Sequencing Data for Improved Diagnostics"
2024	European Human Genetics Conference, Berlin, Germany: "TRAPPC6B Biallelic Variants Cause a Neurodevelopmental Disorder with TRAPP II and Trafficking Disruptions"
2024	European Human Genetics Conference, Berlin, Germany: "Unveiling the Genetic Diversity of Cerebral Palsy Mimics Using Trio-WES in the Turkish Cohort"
2024	European Biotechnology Congress, Leuven, Belgium: "Functional and Structural Landscape Of Cerebral Palsy (Cp) Related Proteins"
2023	148th Annual Meeting American-Neurological-Association (ANA), Philadelphia, PA: "Human Cerebral Organoids with PIDD1 Mutations Implicate AKT-mTOR Pathway Hypoactivity in Lissencephaly"
2023	International Conference on Rare and Undiagnosed Diseases, Tbilisi, Georgia: "Obstacles and Expectations of Rare Disease Patients and Their Families in Türkiye: ISTisNA Project Survey Results"
2021	Annual Meeting of the American-Society-of-Tropical-Medicine-and-Hygiene (ASTMH), Electr Network: "Comparative Phylodynamics of Sars-Cov-2 Variants in the Northeast United States"
2020	53rd European Society of Human Genetics (ESHG) Conference, Electr Network: "Mutation Spectrum of Congenital Heart Disease in 73 Consanguineous Turkish Families"
2020	Annual Meeting of the Congress-of-Neurological-Surgeons, Electr Network: "Integrative Genomics Implicates Genetic Disruption of Prenatal Neurogenesis in Congenital Hydrocephalus"
2020	25th Virtual Annual Scientific Meeting and Education Day of the Society-for-Neuro-Oncology (SNO), Electr Network: "Associations of Genomic Subgroup With Recurrence In Low-Grade Meningiomas"
2020	32nd EORTC-NCI-AACR Symposium on Molecular Targets and Cancer Therapeutics, Electr Network: "Mutations and Copy Number Alterations in Diffuse Gliomas are Shaped by Different Mechanisms"
2020	Annual Scientific Meeting of the American-Association-of-Neurological-Surgeons (AANS), Boston, MA: "IDH-Mutant Gliomas Differ in Distribution of Mitochondrial Genomic Alterations"
2019	68th Annual Meeting of the American-Society-for-Tropical-Medicine-and-Hygiene, National Harbor, MD: "A Deep Sequencing Approach To Define Benzimidazole Resistance Gene Frequencies In Human Hookworm Egg Samples From Kpandai District, Ghana"
2019	24th Annual Scientific Meeting and Education Day of the Society-for-Neuro-Oncology (SNO) / 3rd SNO-SCIDOT Joint Conference on Therapeutic Delivery to the CNS: "Meningioma with Multiple Drivers: Genomic Landscape And Clinical Correlations"

2019	Annual Meeting of the Congress-of-Neurological-Surgeons, San Francisco, CA: "Identification of Peptidyl-Prolyl Cis-Trans Isomerase-Like 4 as a Disease Causing Gene in Intracranial Aneurysms and its Role in Vertebrate CNS Specific Angiogenesis"
2019	Annual Meeting of the Congress-of-Neurological-Surgeons, San Francisco, CA: "Exome Sequencing Defines the Molecular Pathogenesis of Vein of Galen Malformation"
2019	51st Conference of the European-Society-of-Human-Genetics (ESHG) in conjunction with the European Meeting on Psychosocial Aspects of Genetics (EMPAG), Milan, Italy: "Severe speech delay in Cohen Syndrome: three novel mutations and the long-term follow-up of nine patients"
2018	47th Annual Meeting of the Child-Neurology-Society (CNS), Chicago, IL: "Damaging Genomic Variants Constitute a Major Risk Factor for Cerebral Palsy"
2018	107th Annual Meeting of the United-States-and-Canadian-Academy-of-Pathology (USCAP), Vancouver, Canada: "Whole Genome Sequencing of Matched Prostate Cancer and High-Grade Prostatic Intraepithelial Neoplasia Demonstrates Both Shared and Private Mutations"
2018	American Society of Human Genetics Meeting, San Diego, CA: "Cerebral Palsy Gene Discovery Based on WES of Multiplex Consanguineous Families"
2018	Congress of Neurological Surgeons Meeting, Houston, TX: "Meningioma with Multiple Drivers: Genomic Landscape and Clinical Correlations"
2017	5th Quadrennial Meeting of the World-Federation-of-Neuro-Oncology-Societies (WFNOS), Zurich, Switzerland: "Significantly Mutated Genes For Radiation-Associated Meningioma"
2017	American Association of Neurological Surgeons Meeting, Los Angeles, CA: "Clinical and Molecular Features of Genomic Subgroups in Meningioma"
2017	American Society of Human Genetics Meeting, Orlando, FL: "Discovery of cerebral palsy genes through trio-based whole exome sequencing in cryptogenic individuals"
2016	European Biotechnology Congress, Riga, Latvia: "Constitutional mismatch repair defect syndrome: New insights from whole exome sequencing data and functional studies"
2016	Advances in Genome Biology and Technology Meeting, Orlando, FL: "Improvement for whole-exome capture and sequencing using SeqCap EZ MedExome target enrichment kit from Roche/Nimblegen"
2014	Joint Congress of European Neurology, Istanbul, Turkey: "The distinct genetic pattern of ALS in Turkey"
2014	Annual meeting of the Association for Research in Vision and Ophthalmology (ARVO), Orlando, FL: "Next-Generation Exome Sequencing of Primary Congenital Glaucoma (PCG) Families"
2014	Annual meeting of the Association for Research in Vision and Ophthalmology (ARVO), Orlando, FL: "Exome Sequencing of Familial and Sporadic Cases with Pseudoexfoliation Syndrome"

2014 The European Human Genetics Conference, Milan, Italy: "A new autosomal recessive syndrome: Severe delay and dysmorphic features causing by a missense mutation in FTO gene"

2014 American Association of Neurological Surgeons Meeting, San Francisco, CA: "Familial Occurrence of Brain Arteriovenous Malformation: A Novel ACVRL1 Mutation Detected by Whole Exome Sequencing"

2014 Congress of Neurological Surgeons Meeting, Boston, MA: "Whole exome sequencing of sporadic cerebellar hemangioblastoma"

2014 Congress of Neurological Surgeons Meeting, Boston, MA: "Neurosurgical location predicts mutation profile in meningiomas"

2013 4th Quadrennial Meeting of the World-Federation-of-Neuro-Oncology (WFNO) held in conjunction with the 18th Annual Meeting of the Society-for-Neuro-Oncology (SNO), San Francisco, CA: "Genomic Analysis Of Traf7-Only Mutant Meningiomas Reveals Novel Driver Mutations"

2013 4th Quadrennial Meeting of the World-Federation-of-Neuro-Oncology (WFNO) held in conjunction with the 18th Annual Meeting of the Society-for-Neuro-Oncology (SNO), San Francisco, CA: "Correlation Of Genetic Signatures And Histological Subtypes Of Meningiomas"

2013 17th International Congress of Parkinson's Disease and Movement Disorders, Sydney, Australia: "FBXO7 mutation: Phenotypic variability from chorea to early-onset asymmetric parkinsonism within a family"

2013 European Paediatric Neurology Society Congress, Brussels, Belgium: "A New Variant Detected in GRIN2A by Whole-Exome Sequencing in a Patient with Intellectual Disability with Intractable Epilepsy"

2013 European Paediatric Neurology Society Congress, Brussels, Belgium: "Mutations detected in primer microcephaly genes by whole-exome sequencing"

2013 The European Human Genetics Conference, Paris, France: "Autosomal recessive spastic quadriplegia caused by AP4M1 and AP4B1 gene mutations: expansion of the facial and cranial MRI features"

2013 Europediatrics Conference, Glasgow, UK: "Whole-exome sequencing detected CEP135 Mutation: second report in the literature"

2013 Neurometabolic Dysmorphology Symposium, Istanbul, Turkey: "Genetic diagnosis in Joubert syndrome using whole exome sequencing"

2013 Neurometabolic Dysmorphology Symposium, Istanbul, Turkey: "Identification of KIF22 mutation in a patient with short stature, scoliosis and broad nose with whole exome sequencing"

2013 Neurometabolic Dysmorphology Symposium, Istanbul, Turkey: "Co-occurrence of hyperandrogenism and Pakistani type spondyloepiphyseal dysplasia"

2013 CV/SNIS Meeting, Honolulu, HI: "CCM3 is involved in neural-endothelial cell communication in the CNS"

2013 CV/SNIS Meeting, Honolulu, HI: "Identification of intracranial aneurysm genetic susceptibility loci through genome wide association studies"

2012 The European Human Genetics Conference, Nürnberg, Germany: "Eleven patients with Camptodactyly-Arthropathy syndrome in a kindred Turkish family caused by homozygous deletion in PRG4 gene"

2012 Congress of Neurological Surgeons Meeting, Chicago, IL: "Recessive LAMC3 mutations cause malformations of occipital cortical development"

2012 Annual Meeting of European Academy of Childhood Disability, Istanbul, Turkey: "A first report of TMCO1 defect syndrome in a non-Amish patient"

2012 Annual Meeting of European Academy of Childhood Disability, Istanbul, Turkey: "A new Turkish case with Ataxia-Oculomotor Apraxia syndrome type 1 diagnosed by whole-exome sequencing"

2012 American Society of Human Genetics Meeting, San Francisco, CA: "Identification of a novel missense mutation in RAD51 in a large family with congenital mirror movements"

2010 The European Human Genetics Conference, Gothenburg, Sweden: "Targeted next generation sequencing identifies a mutation associated with cerebellar hypoplasia and mental retardation with quadrupedal locomotion"

2010 The European Human Genetics Conference, Gothenburg, Sweden: "Heterozygous 5p13.3-13.2 deletion in a patient with type I Chiari malformation and bilateral Duane retraction syndrome"

2010 American Society of Human Genetics Meeting, Washington D.C.: "Genome-wide association study of intracranial aneurysm identifies new loci"

2010 Congress of Neurological Surgeons Meeting, San Francisco, CA: "Copy Number Variation Analysis of Malformation of Cortical Development for Candidate Gene Identification Using High Density SNP Microarray"

2010 Congress of Neurological Surgeons Meeting, San Francisco, CA: "Genome-Wide Analysis of Copy Number Variations in Intracranial Aneurysm"

2010 Congress of Neurological Surgeons Meeting, San Francisco, CA: "Genome-Wide Association Study Identifies Five Risk Lock for Intracranial Aneurysms"

2010 Congress of Neurological Surgeons Meeting, San Francisco, CA: "Mutations within the PLA2G6 Gene Cause Infantile Neuroaxonal Dystrophy"

2010 Congress of Neurological Surgeons Meeting, San Francisco, CA: "Whole Exome Sequencing in a Patient with Malformation of Cortical Development Identifies a Novel Gene Involved in Brain Development"

2010 American Academy of Child and Adolescent Psychiatry Annual Meeting, New York, NT: "A Project to Identify Genes Contributing to Autism in Consanguineous Egyptian Pedigrees by Utilizing Homozygosity Mapping and Whole-Exome Sequencing"

2009 Congress of Neurological Surgeons Meeting, New Orleans, LA: "Mutations in the Type IV Collagens, COL4A1 and COL4A2 are Associated with Intraventricular Hemorrhage in Preterm Infants"

2009 American Society of Human Genetics Meeting, Honolulu, HI: "A Novel Form of Recessive Cerebellar Atrophy Links To 5q31.1-q33.1"

2009 American Society of Human Genetics Meeting, Honolulu, HI: "Novel VLDLR Microdeletion Identified in Two Turkish Siblings with Pachygyria and Pontocerebellar Atrophy"

2009 American Society of Human Genetics Meeting, Honolulu, HI: "Homozygosity mapping in patients with malformations of cortical development"

2009 American Society of Human Genetics Meeting, Honolulu, HI: "Copy number variation analysis in patients with malformations of cortical development"

2009 American Society of Human Genetics Meeting, Honolulu, HI: "Developmental changes in human neocortical transcriptome revealed by RNA-seq"

2009 American Stroke Association International Stroke Conference, The Need to Know: Key Clinical Research and Trials (Late-Breaking Science), San Diego, CA: "Whole Genome Association of Intracranial Aneurysm Identifies Susceptibility Loci"

2008 The European Human Genetics Conference, Barcelona, Spain: "A second candidate gene causing aniridia: preliminary results"

2008 Congress of Neurological Surgeons Meeting, Orlando, FL: "Copy Number Variant Analysis of Patients with Malformations of Cortical Development"

2008 Scientific Congress of Turkish Neurosurgical Society, Antalya, Turkey: "Co-occurrence of familial neurofibromatosis type I and Knobloch syndrome"

2008 Days of Molecular Medicine, Cognitive Dysfunction in Disease: Mechanisms and Therapies, Stockholm, Sweden: "Complete sequencing of the human brain transcriptome"

2008 American Stroke Association International Stroke Conference, New Orleans, LA: "Genome Wide Association Study of Intracranial Aneurysms in the Finnish Population"

2008 American Stroke Association International Stroke Conference, New Orleans, LA: "High Resolution Copy Number Variation Analysis of Sporadic and Familial CCM Patients"

2007 Congress of Neurological Surgeons Meeting, San Diego, CA: "Deletion within CCM2 Interval Causes A Novel Syndrome of Cerebral Cavernous Malformation and Greig Cephalopolysyndactyly"

2007 Congress of Neurological Surgeons Meeting, San Diego, CA: "Linkage Analysis Identifies A Novel Locus for MRI-Negative Generalized Tonic Clonic Epilepsy"

2007 Congress of Neurological Surgeons Meeting, San Diego, CA: "Linkage and Copy Number Variation Analysis of Large Families and Sibling Pairs Demonstrates Locus Heterogeneity for Familial Intracranial Aneurysms"

2007 Congress of Neurological Surgeons Meeting, San Diego, CA: "Rapid Identification of Disease-Causing Mutation in A Family with Autosomal Recessive Parkinson Disease Using Copy Number Analysis within Linkage Interval"

2006 American Association of Neurological Surgeons Meeting, San Francisco, CA: "Lack of TGFBR Mutations in Intracranial Aneurysm"

2006 American Association of Neurological Surgeons Meeting, San Francisco, CA: "Genetic Heterogeneity of Intracranial Aneurysms"

2006 American Association of Neurological Surgeons Meeting, San Francisco, CA: "Linkage of Intracranial Aneurysm Susceptibility Genes to Multiple Loci throughout the Human Genome"

2006 American Association of Neurological Surgeons Meeting, San Francisco, CA: "Cerebral venous malformations have distinct genetic origin from cerebral cavernous malformations"

2006	Congress of Neurological Surgeons Meeting. Chicago, IL: "The Yield of Radiological Screening of At-Risk Asymptomatic Members in Large Intracranial Aneurysm Families"
2006	Congress of Neurological Surgeons Meeting. Chicago, IL: "Molecular Genetic Analysis of 2 Large Kindred with Intracranial Aneurysms Demonstrates Linkage to 11q24-25 and 14q23-31"
2006	Congress of Neurological Surgeons Meeting. Chicago, IL: "Lack of Association between Type I or Type II Transforming Growth Factor Receptor Mutations and Familial Forms of Intracranial Aneurysm"
2004	Scientific Congress of Turkish Neurosurgical Society, Antalya, Turkey: "Investigation of neuro protective effects of a caspase 3 Inhibitor AC-DMQD-CHO in experimental spinal cord trauma model in rats"
2004	Scientific Congress of Turkish Neurosurgical Society, Antalya, Turkey: "Investigation of neuro protective effects of a caspase 1 Inhibitor AC-YVAD-CMK in experimental spinal cord trauma model in rats"
2004	Scientific Congress of Turkish Neurosurgical Society, Antalya, Turkey: "Investigation of neuro protective effects of a calpain 6 inhibitor SJA6017 in experimental spinal cord trauma model in rats"
2004	National Congress of Prenatal Diagnosis and Medical Genetics, Antalya, Turkey: "Factor V A4070G, MTHFR A1209C and PAI-1 4G/5G Polymorphisms in Complicated Pregnancies of Unknown Etiology"
2002	National Prenatal Diagnosis and Medical Genetics Congress, Konya, Turkey: "Methylenetetrahydrofolate reductase Gene polymorphisms in End-Stage Renal Failure Patients"

Professional Service:

Peer Review Groups/Grant Study Sections:

2025	Member, Special Emphasis Panel/Scientific Review Group, ZNS1 SRB-V(15), NIH/NINDS
2024	Member, Special Emphasis Panel/Scientific Review Group, ZNS1 SRB-S (26) NINDS Human Genetics Resource Center Contract Review, NIH/NINDS
2022	Member, Special Emphasis Panel, ZDE1 YM 12 U01, Clinical Studies, NIH/NIDCR
2020	Member, Special Emphasis Panel, ZHG1 HGR-P (O1) COVID Supplements, NIH/NHGRI
2019	Ad hoc member, Center for Inherited Disease Research Access Committee (CIDR) 01, NIH/NHGRI
2017	Member, Special Emphasis Panel ZHG1 HGR-P J1, Diversity Action Plan, NIH/NHGRI
2016	Member, Special Emphasis Panel ZHG1 HGR-P O1, Gabriella Miller Kids First Sequencing Center Review, NIH/NHGRI
2016	Member, Special Emphasis Panel ZHG1 HGR-P M1, Center for Inherited Disease Research (CIDR) Contract Review, NIH/NHGRI

Journal Service:

2010-present	Reviewer for *Journal of Medical Genetics, Clinical Genetics, Stroke, Neurosurgery, Cerebrovascular Diseases, American Journal of Human Genetics*

Professional Service for Professional Organizations:

2018-present	Member, AACR Project GENIE Consortium, Genomics & Analytical Working Group
2017-2021	Member, Centers for Mendelian Genomics and Knock Out Mouse Project Joint Working Committee, National Human Genome Research Institute, NIH
2017	Member, Election Committee, Aziz Sancar Science and Service Awards, Health Institutes of Turkey
2015-2021	Member, Steering Committee, Centers for Mendelian Genomics, National Human Genome Research Institute, NIH
2012-2021	Member, IRB/Consent Review working group, Centers for Mendelian Genomics, National Human Genome Research Institute, NIH

University Service:

Medical School/University Committees, Acibadem University:

2024-present	Member, ATADEK Medical Research Ethics Committee
2024-present	Member, Steering Committee, Institute of Health Sciences
2024-present	Member, Steering Committee, School of Medicine
2024-present	Member, International Students Admissions Committee, School of Medicine
2023-present	Member, Steering Committee, ACURARE Rare Diseases and Orphan Drugs Application and Research Center
2023-present	Member, MD/PhD Program Admissions Committee
2022-present	Member, International Visibility and Recognition Committee
2022-2023	Member, MD/PhD Program Development Working Group
2021-present	Member, Ethics Committee, ACU Biobank Unit
2021-present	Member, Ethics Committee, ACURARE Rare Diseases and Orphan Drugs Application and Research Center

Medical School/Hospital Committees, Yale University:

2020-2021	Member, Yale SARS-CoV-2 Genomic Surveillance Initiative
2019-2021	Member, Genomic Cancer Profiling Working Group, Comprehensive Cancer Center
2018-2021	Member, Graduate Admissions Committee, MCGD Track
2014-2016	Member, Cancer Genomics Committee

Departmental Committees, Acibadem University:

2022-present	Member, Admissions Committee, Translational Medicine Program, Institute of Health Sciences
2021-present	Member, Admissions Committee, Biostatistics and Bioinformatics Program, Institute of Health Sciences
2021-present	Member, Admissions Committee, Genome Studies Program, Institute of Health Sciences

Departmental Committees, Yale University:

2018-2021	Member, Mentoring Committee, Department of Genetics

Mentored Thesis:

Zhang, Ce	2019-2024, Human Cerebral Organoids for Modelin Neurodevelopmental and Infectious Disease
Bulut, Aybike	2021-2023, Identification and Characterization of in Early Onset Multisystemic Neurodegeneration
Yigit, Ayca	2021-2023, Utilization of Whole and Clinical Exome Sequencing Data in a Turkish Cohort of Cerebral Palsy Patients to Identify Novel Candidate Genes and Variants and Increase Diagnosis
Cetinkaya, Gulinay	2022-2023, Variant Interpretation and Prioritization of Early-Onset Schizophrenia Patients using Whole Exome Sequencing
Eksi, Neslihan	2022-2023, Gene Identification in Microcephaly Using Whole-exome Sequencing
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Kupeli, Derya	2023-2024, Whole-Exome Sequencing Analysis Of A Turkish Family With Joint Hypermobility Syndrome
Koroglu, Merve Nur	2024-present, Common Disease Rare Variant: An Analysis Framework for Exploring Genetic Architecture of Complex Diseases
Aral, Melisa	2024-present, Investigation of Genetic Causation Underlying Microlissencephaly Spectrum of Disorders
Kaya, Pamir	2024-present, Gene Idenfication in Microcephaly Families
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(*Equal contribution)

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