Assoc. Prof. KAYA BİLGÜVAR

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Education Information

Doctorate, Marmara University, Institute Of Health Sciences, Turkey 2002 - 2022
Undergraduate, Marmara University, School Of Medicine, Turkey 1994 - 2000

Foreign Languages

German, A1 Beginner
English, C2 Mastery
Turkish, C2 Mastery

Research Areas

Biological Information, Genetic Disorders, Genomics, Moleculer Biology of Cancer, Biopsychology, Population Genetics, Fundamental Medical Sciences

Academic Titles / Tasks

Assistant Professor, Acibadem Mehmet Ali Aydinlar University, School Of Medicine, Department Of Medical Sciences, 2023 - Continues
Associate Professor, Yale University, School of Medicine, Neurosurgery, 2021 - Continues
Associate Professor, Yale University, School of Medicine, Genetics, 2021 - Continues
Expert PhD, Acibadem Mehmet Ali Aydinlar University, Additional Departments, 2021 - 2024
Associate Professor, Yale University, School of Medicine, Neurosurgery, 2020 - 2021
Associate Professor, Yale University, School of Medicine, Genetics, 2019 - 2021
Assistant Professor, Yale University, School of Medicine, Genetics, 2013 - 2019
Researcher, Yale University, School of Medicine, Neurosurgery, 2007 - 2013
Other, Yale University, School of Medicine, Neurosurgery, 2004 - 2007

Published journal articles indexed by SCI, SSCI, and AHCI

I. **Reply to Pisan et al.: Pathogenicity of inherited TRAF7 mutations in congenital heart disease**
Mishra-Gorur K., Barak T., Kaulen L. D., Henegariu O., Jin S. C., Aguilera S. M., Yalbir E., Goles G., Nishimura S.,

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II. **TRAPPC6B biallelic variants cause a neurodevelopmental disorder with TRAPP II and trafficking disruptions**
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III. **Super-enhancer hijacking drives ectopic expression of hedgehog pathway ligands in meningiomas**
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IV. **Toward Precision Oncology in Glioblastoma with a Personalized Cancer Genome Reporting Tool and Genetic Changes Identified by Whole Exome Sequencing**
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V. **Biallelic frameshift variants in PHLDB1 cause mild-type osteogenesis imperfecta with regressive spondylometaphyseal changes**
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VI. **Pleiotropic role of TRAF7 in skull-base meningiomas and congenital heart disease**
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VII. **Inborn errors of OAS-RNase L in SARS-CoV-2-related multisystem inflammatory syndrome in children**
Lee D., Le Pen J., Yatim A., Dong B., Aquino Y., Ogishi M., Pescarmona R., Talouarn E., Rinchai D., Zhang P., et al.
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VIII. **LRRC23 truncation impairs radial spoke 3 head assembly and sperm motility underlying male infertility**
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IX. **Combining genomic and epidemiological data to compare the transmissibility of SARS-CoV-2 variants Alpha and Iota**
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X. **Application of multiplex amplicon deep-sequencing (MAD-seq) to screen for putative drug resistance markers in the Necator americanus isotype-1 β-tubulin gene**
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XII. **Mutation spectrum of congenital heart disease in a consanguineous Turkish population**
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Earnest R., Uddin R., Matluk N., Renzette N., Turbett S. E., Siddle K. J., Loreth C., Adams G., Tomkins-Tinch C. H., Petrone M. E., et al.
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XVI. **D-bifunctional protein deficiency caused by splicing variants in a neonate with severe peroxisomal dysfunction and persistent hypoglycemia**
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Calame D. G., Bakhtiari S., Logan R., Coban-Akdemir Z., Du H., Mitani T., Fatih J. M., Hunter J. V., Herman I., Pehlivan D., et al.
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XVIII. **PPIL4 is essential for brain angiogenesis and implicated in intracranial aneurysms in humans**
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XIX. **Bi-allelic variants in SPATA5L1 lead to intellectual disability, spastic-dystonic cerebral palsy, epilepsy, and hearing loss**
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XX. **Erratum to: Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia.**
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XXI. **The genetic structure of the Turkish population reveals high levels of variation and admixture**
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XXII. **Mutation in ZDHHC15 Leads to Hypotonic Cerebral Palsy, Autism, Epilepsy, and Intellectual Disability**
Lewis S. A., Bakhtiari S., Heim J., Cornejo P., Liu J., Huang A., Musmacker A., Jin S. C., Bilguvar K., Padilla-Lopez S. R., et al.
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Alsharhan H., He M., Edmondson A. C., Daniel E. J. P., Chen J., Donald T., Bakhtiari S., Amor D. J., Jones E. A., Vassallo G., et al.
JOURNAL OF INHERITED METABOLIC DISEASE, no.4, pp.1001-1012, 2021 (SCI-Expanded)

XXVI. **Resolution of sclerotic lesions of dysosteosclerosis due to biallelic SLC29A3 variant in a Turkish girl**
ULUDAĞ ALKAYA D., Akpinar E., Bilguvar K., Tuysuz B.
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XXVII. **Recessive COL4A2 Mutation Leads to Intellectual Disability, Epilepsy, and Spastic Cerebral Palsy**
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XXVIII. **Associations of meningioma molecular subgroup and tumor recurrence.**
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XXIX. **Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia**
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XXX. **Genetic Defects in DNAH2 Underlie Male Infertility With Multiple Morphological Abnormalities of the Sperm Flagella in Humans and Mice**
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XXXI. **Integrated mutational landscape analysis of uterine leiomyosarcomas**
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XXXII. **Sequential filtering for clinically relevant variants as a method for clinical interpretation of whole exome sequencing findings in glioma**
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XXXIII. **METAP1mutation is a novel candidate for autosomal recessive intellectual disability**
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XXXIV. **Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome**
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XXXV. **Neuroinvasion of SARS-CoV-2 in human and mouse brain**
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JOURNAL OF EXPERIMENTAL MEDICINE, vol.218, no.3, 2021 (SCI-Expanded)

XXXVI. **A patient with mental retardation, enteropathy, deafness, peripheral neuropathy, ichthyosis, keratodermia syndrome caused by AP1B1 gene variant**
Meric R., Ercan-Sencicek A. G., ULUDAĞ ALKAYA D., Sahin Y., Sar M., Bilguvar K., Tuysuz B.
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XXXVII. **Integrative Genomics Implicates Genetic Disruption of Prenatal Neurogenesis in Congenital Hydrocephalus**
Panchagnula S., Jin S. C., Dong W., Kundishora A., Moreno-De-Luca A., Furey C. G., Allocco A. A., Walker R., Nelson-Williams C., Smith H., et al.
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XXXVIII. **Mutation spectrum of congenital heart disease in 73consanguineous Turkish families**
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EUROPEAN JOURNAL OF HUMAN GENETICS, no.SUPPL 1, pp.263-264, 2020 (SCI-Expanded)

XXXIX. **Mutations and Copy Number Alterations in IDH Wild-Type Glioblastomas Are Shaped by Different Oncogenic Mechanisms**
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XL. **Correlations between genomic subgroup and clinical features in a cohort of more than 3000 meningiomas**
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XLII. **Inborn errors of type I IFN immunity in patients with life-threatening COVID-19**
Zhang Q., Bastard P., Liu Z., Le Pen J., Moncada-Velez M., Chen J., Ogishi M., Sabli I. K. D., Hodeib S., Korol C., et al.
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XLVI. **Whole exome sequencing-based analysis to identify DNA damage repair deficiency as a major contributor to gliomagenesis in adult diffuse gliomas**
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XLVII. **Bi-allelic GAD1 variants cause a neonatal onset syndromic developmental and epileptic encephalopathy**
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XLIX. **Mutations in Chromatin Modifier and Ephrin Signaling Genes in Vein of Galen Malformation**
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L. **Whole-exome sequencing of cervical carcinomas identifies activating ERBB2 and PIK3CA mutations as targets for combination therapy**
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LII. **Loss of Protocadherin-12 Leads to Diencephalic-Mesencephalic Junction Dysplasia Syndrome**
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LIII. **De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias**
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LIV. **Meningioma With Multiple Drivers: Genomic Landscape and Clinical Correlations**
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LVI. **De Novo Mutation in Genes Regulating Neural Stem Cell Fate in Human Congenital Hydrocephalus**
Furey C. G., Choi J., Jin S. C., Zeng X., Timberlake A. T., Nelson-Williams C., Mansuri M. S., Lu Q., Duran D., Panchagnula S., et al.
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LVII. **Integrated genomic analyses of de novo pathways underlying atypical meningiomas.**
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Yilmaz S., ULUDAĞ ALKAYA D., KASAPÇOPUR Ö., BARUT K., Akdemir E. S., Celen C., Youngblood M. W., Yasuno K., Bilguvar K., Gunel M., et al.
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LIX. **Homozygous CAPN1 mutations causing a spastic-ataxia phenotype in 2 families**
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LX. **Contribution of rare inherited and de novo variants in 2,871 congenital heart disease probands**
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LXI. **AAV-mediated direct in vivo CRISPR screen identifies functional suppressors in glioblastoma**
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LXII. **GABBR2 Mutations Determine Phenotype in Rett Syndrome and Epileptic Encephalopathy**
Yoo Y., Jung J., Lee Y., Lee Y., Cho H., Na E., Hong J., Kim E., Lee J. S., Lee J. S., et al.
ANNALS OF NEUROLOGY, vol.82, no.3, pp.466-478, 2017 (SCI-Expanded)

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LXV. **Integrated genomic analyses of de novo pathways underlying atypical meningiomas**
Harmanci A. S., Youngblood M. W., Clark V. E., Coskun S., Henegariu O., Duran D., Erson-Omay E. Z., Kaulen L. D., Lee T. I., Abraham B. J., et al.
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