

## Doç. Dr. KAYA BİLGÜVAR

### Kişisel Bilgiler

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Publons / Web Of Science ResearcherID: EOE-1842-2022

ScopusID: 9037414300

### Eğitim Bilgileri

Doktora, Marmara Üniversitesi, Sağlık Bilimleri Enstitüsü, Türkiye 2002 - 2022

Lisans, Marmara Üniversitesi, Tıp Fakültesi, Türkiye 1994 - 2000

### Yabancı Diller

İngilizce, C2 Ustalık

Türkçe, C2 Ustalık

Almanca, A1 Başlangıç

### Araştırma Alanları

Biyoenformasyon, Genetik Bozuklukların Moleküler Biyolojisi, Genomiks, Kanser Moleküler Biyolojisi, Biyopsikoloji, Populasyon Genetiği, Temel Tıp Bilimleri

### Akademik Unvanlar / Görevler

Dr. Öğr. Üyesi, Acibadem Mehmet Ali Aydınlar Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2023 - Devam Ediyor

Doç. Dr., Yale University, School of Medicine, Neurosurgery, 2021 - Devam Ediyor

Doç. Dr., Yale University, School of Medicine, Genetics, 2021 - Devam Ediyor

Uzman Dr., Acibadem Mehmet Ali Aydınlar Üniversitesi, Rektörlüğe Bağlı Bölümler, 2021 - 2024

Doç. Dr., Yale University, School of Medicine, Neurosurgery, 2020 - 2021

Doç. Dr., Yale University, School of Medicine, Genetics, 2019 - 2021

Dr. Öğr. Üyesi, Yale University, School of Medicine, Genetics, 2013 - 2019

Araştırmacı, Yale University, School of Medicine, Neurosurgery, 2007 - 2013

Diğer, Yale University, School of Medicine, Neurosurgery, 2004 - 2007

### SCI, SSCI ve AHCI İndekslerine Giren Dergilerde Yayınlanan Makaleler

- Rapid genome sequencing for critically ill infants: an inaugural pilot study from Turkey.**  
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- II. **Reply to Pisan et al.: Pathogenicity of inherited TRAF7 mutations in congenital heart disease**  
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- III. **TRAPPC6B biallelic variants cause a neurodevelopmental disorder with TRAPP II and trafficking disruptions**  
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- V. **Toward Precision Oncology in Glioblastoma with a Personalized Cancer Genome Reporting Tool and Genetic Changes Identified by Whole Exome Sequencing**  
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- VI. **Biallelic frameshift variants in *PHLDB1* cause mild-type osteogenesis imperfecta with regressive spondylometaphyseal changes**  
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- VII. **Pleiotropic role of TRAF7 in skull-base meningiomas and congenital heart disease**  
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- VIII. **Inborn errors of OAS-RNase L in SARS-CoV-2-related multisystem inflammatory syndrome in children**  
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- IX. **LRR23 truncation impairs radial spoke 3 head assembly and sperm motility underlying male infertility**  
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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  $\beta$ -tubulin gene**  
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- XI. **Combining genomic and epidemiological data to compare the transmissibility of SARS-CoV-2 variants Alpha and Iota**  
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- XII. **Biallelic *BICD2* variant is a novel candidate for Cohen-like syndrome**  
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- XIII. **Mutation spectrum of congenital heart disease in a consanguineous Turkish population**  
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- XIV. **The risk of COVID-19 death is much greater and age dependent with type I IFN autoantibodies**  
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- XV. **Further delineation of familial polycystic ovary syndrome (PCOS) via whole-exome sequencing: PCOS-related rare *FBN3* and *FN1* gene variants are identified**  
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- XVI. **Comparative transmissibility of SARS-CoV-2 variants Delta and Alpha in New England, USA**  
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- XVIII. **Biallelic loss-of-function variants in the splicing regulator *NSRP1* cause a severe neurodevelopmental disorder with spastic cerebral palsy and epilepsy**  
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- XIX. ***PPIL4* is essential for brain angiogenesis and implicated in intracranial aneurysms in humans**  
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- XXI. **Erratum to: Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia.**  
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- XXIV. **Autoantibodies neutralizing type I IFNs are present in ~4% of uninfected individuals over 70 years old and account for ~20% of COVID-19 deaths**

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- XXV. **X-linked recessive TLR7 deficiency in ~1% of men under 60 years old with life-threatening COVID-19**  
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- XXIX. **Associations of meningioma molecular subgroup and tumor recurrence.**  
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- XXX. **Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia**  
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- XXXI. **Genetic Defects in DNAH2 Underlie Male Infertility With Multiple Morphological Abnormalities of the Sperm Flagella in Humans and Mice**  
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- XXXIII. **Sequential filtering for clinically relevant variants as a method for clinical interpretation of whole exome sequencing findings in glioma**  
Ülgen E., Can Ö., Bilguvar K., Akyerli Boylu C., Kılıçturgay Yüksel Ş., Erşen Danyeli A., Sezerman O. U., Yakıcıer M. C., Pamir M. N., Özduman K.  
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- XXXIV. **METAP1 mutation is a novel candidate for autosomal recessive intellectual disability**  
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- XXXV. **Neuroinvasion of SARS-CoV-2 in human and mouse brain**  
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- XXXVI. **Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome**  
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- XXXVII. **A patient with mental retardation, enteropathy, deafness, peripheral neuropathy, ichthyosis, keratoderma syndrome caused by <i>AP1B1</i> gene variant**

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CLINICAL DYSMORPHOLOGY, cilt.30, sa.1, ss.54-57, 2021 (SCI-Expanded)

- XXXVIII. **Mutations and Copy Number Alterations in IDH Wild-Type Glioblastomas Are Shaped by Different Oncogenic Mechanisms**

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- XXXIX. **Mutation spectrum of congenital heart disease in 73 consanguineous Turkish families**

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- XL. **Integrative Genomics Implicates Genetic Disruption of Prenatal Neurogenesis in Congenital Hydrocephalus**

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- XLI. **Correlations between genomic subgroup and clinical features in a cohort of more than 3000 meningiomas**

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- XLII. **Exome sequencing implicates genetic disruption of prenatal neuro-gliogenesis in sporadic congenital hydrocephalus**

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- XLIII. **Inborn errors of type I IFN immunity in patients with life-threatening COVID-19**

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- XLV. **Mutations disrupting neuritogenesis genes confer risk for cerebral palsy**

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- XLVI. **Human CRY1 variants associate with attention deficit/hyperactivity disorder**

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- XLVII. **Bi-allelic <i>GAD1</i> variants cause a neonatal onset syndromic developmental and epileptic encephalopathy**

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- XLVIII. Whole exome sequencing-based analysis to identify DNA damage repair deficiency as a major contributor to gliomagenesis in adult diffuse gliomas**  
 Ulgen E., CAN Ö., Bilguvar K., OKTAY Y., AKYERLİ BOYLU C., Danyeli A., Yakicier M. C., Sezerman O. U., Pamir M. N., Ozduman K.  
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- XLIX. Identification of a dominant MYH11 causal variant in chronic intestinal pseudo-obstruction: Results of whole-exome sequencing**  
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- L. Mutations in Chromatin Modifier and Ephrin Signaling Genes in Vein of Galen Malformation**  
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- LII. Clonal evolution analysis of paired anaplastic and well-differentiated thyroid carcinomas reveals shared common ancestor**  
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- LIII. De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias**  
 Helbig K. L., Lauerer R. J., Bahr J. C., Souza I. A., Myers C. T., Uysal B., Schwarz N., Gandini M. A., Huang S., Keren B., et al.  
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- LIV. Loss of Protocadherin-12 Leads to Diencephalic-Mesencephalic Junction Dysplasia Syndrome**  
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- LV. Meningioma With Multiple Drivers: Genomic Landscape and Clinical Correlations**  
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- LVI. Biallelic loss of human CTNNA2, encoding alpha N-catenin, leads to ARP2/3 complex overactivity and disordered cortical neuronal migration**  
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- LVII. De Novo Mutation in Genes Regulating Neural Stem Cell Fate in Human Congenital Hydrocephalus**  
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- LVIII. Integrated genomic analyses of de novo pathways underlying atypical meningiomas.**  
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- LIX. Genotype-phenotype investigation of 35 patients from 11 unrelated families with camptodactyly-**

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- LX. **Homozygous *CAPN1* mutations causing a spastic-ataxia phenotype in 2 families**  
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- LXI. **Contribution of rare inherited and *de novo* variants in 2,871 congenital heart disease probands**  
Jin S. C., Homsy J., Zaidi S., Lu Q., Morton S., DePalma S. R., Zeng X., Qi H., Chang W., Sierant M. C., et al.  
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Erturk O., Bilguvar K., KORKMAZ M. B., Bayri Y., Bayrakli F., Arlier Z., Ozturk A. K., YALÇINKAYA C., Tuysuz B., State M. W., et al.

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**CXXVIII. A novel heterozygous deletion within the 3' region of the <i>PAX6</i> gene causing isolated aniridia in a large family group**

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## **Diğer Dergilerde Yayınlanan Makaleler**

- I. **Severe Phenotype in Patients with X-linked Hydrocephalus Caused by a Missense Mutation in <i>L1CAM</i>**  
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- II. **<i>ALPK3</i> gene mutation in a patient with congenital cardiomyopathy and dysmorphic features**  
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- III. **Digenic mutations of human OCRL paralogs in Dent's disease type 2 associated with Chiari I malformation**  
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- II. **COMPARATIVE PHYLODYNAMICS OF SARS-COV-2 VARIANTS IN THE NORTHEAST UNITED STATES**  
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- III. **ASSOCIATIONS OF GENOMIC SUBGROUP WITH RECURRENCE IN LOW-GRADE MENINGIOMAS**  
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- IV. **Mutations and copy number alterations in diffuse gliomas are shaped by different mechanisms**  
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- VI. **MENINGIOMA WITH MULTIPLE DRIVERS: GENOMIC LANDSCAPE AND CLINICAL CORRELATIONS**  
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- VII. **Exome Sequencing Defines the Molecular Pathogenesis of Vein of Galen Malformation**  
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- VIII. **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**  
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- IX. **Severe speech delay in Cohen Syndrome: three novel mutations and the long-term follow-up of nine patients**  
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- X. **A DEEP SEQUENCING APPROACH TO DEFINE BENZIMIDAZOLE RESISTANCE GENE FREQUENCIES IN HUMAN HOOKWORM EGG SAMPLES FROM KPANDAI DISTRICT, GHANA**  
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- XI. **Damaging Genomic Variants Constitute a Major Risk Factor for Cerebral Palsy**  
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- XII. **Whole Genome Sequencing of Matched Prostate Cancer and High-Grade Prostatic Intraepithelial Neoplasia Demonstrates Both Shared and Private Mutations**  
Wilson P., Schulz W., Guo X., Bilguvar K., Humphrey P.  
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- XIII. **SIGNIFICANTLY MUTATED GENES FOR RADIATION-ASSOCIATED MENINGIOMA**  
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- XIV. **Clinical and Molecular Features of Genomic Subgroups in Meningioma**  
Youngblood M. W., Clark V., Harmanci A. S., Bai H., Mora D. D., Montejo J., Li C., Zhu H., Erson-Omay E. Z., Bilguvar K., et al.  
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- XVI. **Familial Occurrence of Brain Arteriovenous Malformation: A Novel ACVRL1 Mutation Detected by Whole Exome Sequencing**  
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- XVII. **Somatic V600E *BRAF* mutation causes syringocystadenoma papilliferum**  
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**XVIII. The distinct genetic pattern of ALS in Turkey**

Ozoguz A., Uyan O., Birdal G., Iskender C., Omur O., Lahut S., Agim Z. S., Kartal E., Parman Y., Tan E., et al.  
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**XIX. The distinct genetic pattern of ALS in Turkey**

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**XX. CORRELATION OF GENETIC SIGNATURES AND HISTOLOGICAL SUBTYPES OF MENINGIOMAS**

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**XXI. GENOMIC ANALYSIS OF TRAF7-ONLY MUTANT MENINGIOMAS REVEALS NOVEL DRIVER MUTATIONS**

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**XXII. Identification of a novel missense mutation in RAD51 in a large family with congenital mirror movements**

ONAT O. E., GÜLSÜNER S. İ., BİLGİN AKDENİZ H. R., KILINÇ G. M., Bilguvar K., BOYACI H., DÖRSCHNER K., UYSAL H., Günel M., ÖZÇELİK H. T.

62nd Annual Meeting of the American Society of Human Genetics, ASHG 2012, San-Francisco, Kostarika, 06 Kasım 2012

**XXIII. Mutations in the Type IV Collagens, COL4A1 and COL4A2 are Associated with Intraventricular Hemorrhage in Preterm Infants**

DiLuna M. L., Bilguvar K., Louvi A., Bizzarro M., Bayrakli F., Bayri Y., Bydon M., Schneider K., Duncan C. C., State M., et al.

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**XXIV. Genome wide association study of intracranial aneurysms in the Finnish population**

Bilguvar K., Bayri Y., DiLuna M., Bayrakli F., Mason C. E., Bydon M., Niemela M., Laakso A., Hernesniemi J., Jaaskelainen J. E., et al.

33rd International Stroke Conference, Louisiana, Amerika Birleşik Devletleri, 19 - 21 Şubat 2008, ss.568

**XXV. High resolution copy number variation analysis of sporadic and familial CCM patients**

Bayri Y., Ho W. S., Bilguvar K., DiLuna M., Bydon M., Collins L. A., Bayrakli F., Mason C. E., State M. W., Gunel M.

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**XXVI. Linkage and copy number variation analysis of large families and sibling pairs demonstrates locus heterogeneity for familial intracranial aneurysms**

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57th Annual Congress of Neurological Surgery (CNS 2007), California, Amerika Birleşik Devletleri, 17 - 19 Eylül 2007, ss.198

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## **Metrikler**

Yayın: 177

Atıf (WoS): 14884

Atıf (Scopus): 16507

H-İndeks (WoS): 52

H-İndeks (Scopus): 55