

Doç. Dr. KAYA BİLGÜVAR

Kişisel Bilgiler

E-posta: Kaya.Bilguvar@acibadem.edu.tr

Web: <https://avesis.acibadem.edu.tr/Kaya.Bilguvar>

Uluslararası Araştırmacı ID'leri

ORCID: 0000-0002-7313-7652

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

1. 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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PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA, cilt.121, sa.12, 2024 (SCI-Expanded)

- II. **TRAPPC6B biallelic variants cause a neurodevelopmental disorder with TRAPP II and trafficking disruptions**
Almoussa H., Lewis S. A., Bakhtiari S., Nordlie S. H., Pagnozzi A., Magee H., Efthymiou S., Heim J. A., Cornejo P., Zaki M. S., et al.
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- III. **Super-enhancer hijacking drives ectopic expression of hedgehog pathway ligands in meningiomas**
Youngblood M. W., Erson-Omay Z., Li C., Najem H., Coskun S., Tyrtova E., Montejo J. D., Miyagishima D. F., Barak T., Nishimura S., et al.
NATURE COMMUNICATIONS, cilt.14, sa.1, 2023 (SCI-Expanded)
- IV. **Toward Precision Oncology in Glioblastoma with a Personalized Cancer Genome Reporting Tool and Genetic Changes Identified by Whole Exome Sequencing**
Erdogan O., Ozkaya S. C., ERZİK C., Bilguvar K., ARGA K. Y., Bayraklı F.
OMICS-A JOURNAL OF INTEGRATIVE BIOLOGY, cilt.27, sa.9, ss.426-433, 2023 (SCI-Expanded)
- V. **Biallelic frameshift variants in *PHLDB1* cause mild-type osteogenesis imperfecta with regressive spondylometaphyseal changes**
Tuysuz B., ULUDAĞ ALKAYA D., Geyik F., ALAYLIOĞLU M., Kasap B., KURUĞOĞLU S., Akman Y. E., Vural M., Bilguvar K.
JOURNAL OF MEDICAL GENETICS, cilt.60, sa.8, ss.819-826, 2023 (SCI-Expanded)
- VI. **Pleiotropic role of TRAF7 in skull-base meningiomas and 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., Miyagishima D., et al.
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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**
Hwang J. Y., Chai P., Nawaz S., Choi J., Lopez-Giraldez F., Hussain S., Bilguvar K., Mane S., Lifton R. P., Ahmad W., et al.
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- IX. **Combining genomic and epidemiological data to compare the transmissibility of SARS-CoV-2 variants Alpha and Iota**
Petroni M. E., Rothman J. E., Breban M., Ott I. M., Russell A., Lasek-Nesselquist E., Badr H., Kelly K., Omerza G., Renzette N., et al.
COMMUNICATIONS BIOLOGY, cilt.5, sa.1, 2022 (SCI-Expanded)
- 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**
George S., Suwondo P., Akorli J., Otchere J., Harrison L. M., Bilguvar K., Knight J. R., Humphries D., Wilson M. D., Caccone A., et al.
SCIENTIFIC REPORTS, cilt.12, sa.1, 2022 (SCI-Expanded)
- XI. **Biallelic *BICD2* variant is a novel candidate for Cohen-like syndrome**
ÇAĞLAYAN A. O., Tuysuz B., Gul E., ULUDAĞ ALKAYA D., Yalcinkaya C., Gleeson J. G., Bilguvar K., Gunel M.
JOURNAL OF HUMAN GENETICS, cilt.67, sa.9, ss.553-556, 2022 (SCI-Expanded)
- XII. **Mutation spectrum of congenital heart disease in a consanguineous Turkish population**
Dong W., Kaymakcalan H., Jin S. C., Diab N. S., Tanidir C., Yalcin A. S. Y., Ercan-Sencicek A. G., Mane S., Gunel M., Lifton R. P., et al.
MOLECULAR GENETICS & GENOMIC MEDICINE, cilt.10, sa.6, 2022 (SCI-Expanded)

- XIII. **The risk of COVID-19 death is much greater and age dependent with type I IFN autoantibodies**
Manry J, Bastard P, Gervais A, Le Voyer T, Rosain J, Philippot Q, Michailidis E, Hoffmann H, Eto S, Garcia-Prat M, et al.
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- XIV. **Further delineation of familial polycystic ovary syndrome (PCOS) via whole-exome sequencing: PCOS-related rare *FBN3* and *FN1* gene variants are identified**
KARAKAYA C, Cil A. P, Bilguvar K, Cakir T, Karalok M. H, KARABACAK R. O, ÇAĞLAYAN A. O.
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- XV. **Comparative transmissibility of SARS-CoV-2 variants Delta and Alpha in New England, USA**
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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AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.188, sa.1, ss.357-363, 2022 (SCI-Expanded)
- XVII. ***PPIL4* is essential for brain angiogenesis and implicated in intracranial aneurysms in humans**
Barak T, Ristori E, Ercan-Sencicek A. G., Miyagishima D. F., Nelson-Williams C., Dong W., Jin S. C., Prendergast A., Armero W., Henegariu O., et al.
NATURE MEDICINE, cilt.27, sa.12, ss.2165-2175, 2021 (SCI-Expanded)
- XVIII. **Biallelic loss-of-function variants in the splicing regulator *NSRP1* cause a severe neurodevelopmental disorder with spastic cerebral palsy and epilepsy**
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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- XIX. **Bi-allelic variants in *SPATA5L1* lead to intellectual disability, spastic-dystonic cerebral palsy, epilepsy, and hearing loss**
Richard E. M., Bakhtiari S., Marsh A. P. L., Kaiyrzhanov R., Wagner M., Shetty S., Pagnozzi A., Nordlie S. M., Guida B. S., Cornejo P., et al.
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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**
Kars M. E., Basak A. N., Onat O. E., Bilguvar K., Choi J., Itan Y., Caglar C., Palvadeau R., Casanova J., Cooper D. N., et al.
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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.
NEUROLOGY-GENETICS, cilt.7, sa.4, 2021 (SCI-Expanded)
- XXIII. **X-linked recessive *TLR7* deficiency in ~1% of men under 60 years old with life-threatening COVID-19**
Asano T., Boisson B., Onodi F., Matuozzo D., Moncada-Velez M., Renkilaraj M. R. L. M., Zhang P., Meertens L., Bolze A., Materna M., et al.
SCIENCE IMMUNOLOGY, cilt.6, sa.62, 2021 (SCI-Expanded)
- 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

Bastard P., Gervais A., Le Voyer T., Rosain J., Philippot Q., Manry J., Michailidis E., Hoffmann H., Eto S., Garcia-Prat M., et al.

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- XXV. **Resolution of sclerotic lesions of dysosteosclerosis due to biallelic *SLC29A3* variant in a Turkish girl**
ULUDAĞ ALKAYA D., Akpınar E., Bilguvar K., Tuysuz B.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.185, sa.7, ss.2271-2277, 2021 (SCI-Expanded)
- XXVI. ***ALG13* X-linked intellectual disability: New variants, glycosylation analysis, and expanded phenotypes**
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.
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- XXVII. **Recessive *COL4A2* Mutation Leads to Intellectual Disability, Epilepsy, and Spastic Cerebral Palsy**
Bakhtiari S., Tafakhori A., Jin S. C., Guida B. S., Alehabib E., Firouzbadi S., Bilguvar K., Fahey M. C., Darvish H., Kruer M. C.
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- XXVIII. **Associations of meningioma molecular subgroup and tumor recurrence.**
Youngblood M. W., Miyagishima D. F., Jin L., Gupte T., Li C., Duran D., Montejo J. D., Zhao A., Sheth A., Tyrtova E., et al.
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- XXIX. **Biallelic variants in *HPDL* cause pure and complicated hereditary spastic paraplegia**
Wiessner M., Maroofian R., Ni M., Pedroni A., Muller J. S., Stucka R., Beetz C., Efthymiou S., Santorelli F. M., Alfares A. A., et al.
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- XXX. **Genetic Defects in *DNAH2* Underlie Male Infertility With Multiple Morphological Abnormalities of the Sperm Flagella in Humans and Mice**
Hwang J. Y., Nawaz S., Choi J., Wang H., Hussain S., Nawaz M., Lopez-Giraldez F., Jeong K., Dong W., Oh J., et al.
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- XXXI. **Integrated mutational landscape analysis of uterine leiomyosarcomas**
Choi J., Manzano A., Dong W., Bellone S., Bonazzoli E., Zammataro L., Yao X., Deshpande A., Zaidi S., Guglielmi A., et al.
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- XXXII. ***METAP1* mutation is a novel candidate for autosomal recessive intellectual disability**
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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. **A patient with mental retardation, enteropathy, deafness, peripheral neuropathy, ichthyosis, keratoderma 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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- XXXV. **Neuroinvasion of SARS-CoV-2 in human and mouse brain**
Song E., Zhang C., Israelow B., Lu-Culligan A., Prado A. V., Skriabine S., Lu P., Orr-El Weizman O. W., Liu F., Dai Y., et al.

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- XXXVI. **Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome**
Dyment D. A., O'Donnell-Luria A., Agrawal P. B., Coban Akdemir Z., Aleck K. A., Antaki D., Al Sharhan H., Au P. B., Aydin H., Beggs A. H., et al.

AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.185, sa.1, ss.119-133, 2021 (SCI-Expanded)

- 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.

NEUROSURGERY, ss.195, 2020 (SCI-Expanded)

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

Ulgen E., Karacan S., Gerlevik U., CAN Ö., Bilguvar K., OKTAY Y., B. Akyerli C., K. Yuksel S., ERŞEN DANYELİ A., Tihan T., et al.

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

Dong W., Kaymakcalan H., Diab N., Jin S. C., Tanidir C., Yalcin A., Mane S., Bilguvar K., Brueckner M., Lifton R.

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

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JOURNAL OF NEUROSURGERY, cilt.133, sa.5, ss.1345-1354, 2020 (SCI-Expanded)

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

Jin S. C., Dong W., Kundishora A. J., Panchagnula S., Moreno-De-Luca A., Furey C. G., Allocco A. A., Walker R. L., Nelson-Williams C., Smith H., et al.

NATURE MEDICINE, cilt.26, sa.11, ss.1754-1765, 2020 (SCI-Expanded)

- XLII. **Autoantibodies against type I IFNs in patients with life-threatening COVID-19**

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

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

Jin S. C., Lewis S. A., Bakhtiari S., Zeng X., Sierant M. C., Shetty S., Nordlie S. M., Elie A., Corbett M. A., Norton B. Y., et al.

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

Onat O. E., Kars M. E., GÜL Ş., Bilguvar K., Wu Y., Ozhan A., Aydin C., Basak A. N., Trusso M. A., Goracci A., et al.

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- XLVI. **Bi-allelic *GAD1* variants cause a neonatal onset syndromic developmental and epileptic encephalopathy**

Chatron N., Becker F., Morsy H., Schmidts M., Hardies K., Tuysuz B., Roselli S., Najafi M., ULUDAĞ ALKAYA D., Ashrafzadeh F., et al.

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- XLVII. **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 BOYLUCU C., Danyeli A., Yakicier M. C., Sezerman O. U., Pamir M. N., Ozduman K.

JOURNAL OF NEUROSURGERY, cilt.132, sa.5, ss.1435-1446, 2020 (SCI-Expanded)

- XLVIII. **Identification of a dominant *MYH11* causal variant in chronic intestinal pseudo-obstruction: Results of whole-exome sequencing**
Dong W., Baldwin C., Choi J., Milunsky J. M., Zhang J., Bilguvar K., Lifton R. P., Milunsky A.
CLINICAL GENETICS, cilt.96, sa.5, ss.473-477, 2019 (SCI-Expanded)
- 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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- LI. **Clonal evolution analysis of paired anaplastic and well-differentiated thyroid carcinomas reveals shared common ancestor**
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- LII. **Loss of *Protocadherin-12* Leads to Diencephalic-Mesencephalic Junction Dysplasia Syndrome**
Guemez-Gamboa A., ÇAĞLAYAN A. O., Stanley V., Gregor A., Zaki M. S., Saleem S. N., Musaev D., McEvoy-Venneri J., Belandres D., Akizu N., et al.
ANNALS OF NEUROLOGY, cilt.84, sa.5, ss.638-647, 2018 (SCI-Expanded)
- 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. **Meningioma With Multiple Drivers: Genomic Landscape and Clinical Correlations**
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- LV. **Biallelic loss of human CTNNA2, encoding alpha N-catenin, leads to ARP2/3 complex overactivity and disordered cortical neuronal migration**
Schaffer A. E., Breuss M. W., Caglayan A. O., Al-Sanaa N., Al-Abdulwahed H. Y., Kaymakcalan H., Yilmaz C., Zaki M. S., Rosti R. O., Copeland B., et al.
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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.
NEURON, cilt.99, sa.2, ss.302-318, 2018 (SCI-Expanded)
- LVII. **Integrated genomic analyses of de novo pathways underlying atypical meningiomas.**
Harmanci A. S., Youngblood M. W., Clark V. E., Coşkun S., Henegariu O., Duran D., Erson-Omay E. Z., Kaulen L. D., Lee T. I., Abraham B. J., et al.
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- LVIII. **Genotype-phenotype investigation of 35 patients from 11 unrelated families with camptodactyly-arthropathy-coxa vara-pericarditis (CACP) syndrome**
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.
MOLECULAR GENETICS & GENOMIC MEDICINE, cilt.6, sa.2, ss.230-248, 2018 (SCI-Expanded)

- LIX. **Homozygous *CAPN1* mutations causing a spastic-ataxia phenotype in 2 families**
Kocoglu C., Gundogdu A., Kocaman G., Kahraman-Koytak P., Uluc K., KIZILTAN G., ÇAĞLAYAN A. O., Bilgüv K., Vural A., Basak A. N.
NEUROLOGY-GENETICS, cilt4, sa.1, 2018 (SCI-Expanded)
- LX. **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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- LXI. **AAV-mediated direct *in vivo* CRISPR screen identifies functional suppressors in glioblastoma**
Chow R. D., Guzman C. D., Wang G., Schmidt F., Youngblood M. W., Ye L., Errami Y., Dong M. B., Martinez M. A., Zhang S., et al.
NATURE NEUROSCIENCE, cilt20, sa.10, ss.1329-1341, 2017 (SCI-Expanded)
- 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.
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- LXIII. **Familial occurrence of brain arteriovenous malformation: a novel *ACVRL1* mutation detected by whole exome sequencing**
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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. **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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- VIII. **Exome Sequencing Defines the Molecular Pathogenesis of Vein of Galen Malformation**
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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**
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- XIII. **SIGNIFICANTLY MUTATED GENES FOR RADIATION-ASSOCIATED MENINGIOMA**
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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**
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XIX. The distinct genetic pattern of ALS in Turkey

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XXI. CORRELATION OF GENETIC SIGNATURES AND HISTOLOGICAL SUBTYPES OF MENINGIOMAS

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

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

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

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Metrikler

Yayın: 174

Atf (WoS): 14884

Atf (Scopus): 16501

H-İndeks (WoS): 52

H-İndeks (Scopus): 55