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SCI, SSCI ve AHCI İndekslerine Giren Dergilerde Yayınlanan Makaleler

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.,

Miyagishima D., et al.
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**
Almousa 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.
BRAIN, cilt.147, sa.1, ss.311-324, 2024 (SCI-Expanded)

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., Bayrakli 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.
PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA, cilt.120, sa.16, 2023 (SCI-Expanded)

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.
SCIENCE, cilt.379, sa.6632, ss.554-574, 2023 (SCI-Expanded)

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.
ELIFE, cilt.12, 2023 (SCI-Expanded)

IX. **Combining genomic and epidemiological data to compare the transmissibility of SARS-CoV-2 variants Alpha and Iota**
Petrone 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.
PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA, cilt.119, sa.21, 2022 (SCI-Expanded)

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.
JOURNAL OF OBSTETRICS AND GYNAECOLOGY RESEARCH, cilt.48, sa.5, ss.1202-1211, 2022 (SCI-Expanded)

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.
CELL REPORTS MEDICINE, cilt.3, sa.4, 2022 (SCI-Expanded)

XVI. **D-bifunctional protein deficiency caused by splicing variants in a neonate with severe peroxisomal dysfunction and persistent hypoglycemia**
Werner K. M., Cox A. J., Qian E., Jain P., Ji W., Tikhonova I., Castaldi C., Bilguvar K., Knight J., Ferdinandusse S., et al.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.188, sa.1, ss.357-363, 2022 (SCI-Expanded)

XVII. **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.
GENETICS IN MEDICINE, cilt.23, sa.12, ss.2455-2460, 2021 (SCI-Expanded)

XVIII. **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)

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.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.108, sa.10, ss.2006-2016, 2021 (SCI-Expanded)

XX. **Erratum to: Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia.**
Wiessner M., Maroofian R., Ni M., Pedroni A., Müller J. S., Stucka R., Beetz C., Efthymiou S., Santorelli F. M., Alfares A. A., et al.
Brain : a journal of neurology, cilt.144, sa.8, 2021 (SCI-Expanded)

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.
PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA, cilt.118, sa.36, 2021 (SCI-Expanded)

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. **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.
SCIENCE IMMUNOLOGY, cilt.6, sa.62, 2021 (SCI-Expanded)

XXIV. **X-linked recessive TLR7 deficiency in ~1% of men under 60 years old with life-threatening COVID-**

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)

XXV. **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.

JOURNAL OF INHERITED METABOLIC DISEASE, sa.4, ss.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.

AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.185, sa.7, ss.2271-2277, 2021 (SCI-Expanded)

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.

NEUROLOGY-GENETICS, cilt.7, sa.3, 2021 (SCI-Expanded)

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.

Neuro-oncology, cilt.23, sa.5, ss.783-794, 2021 (SCI-Expanded)

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.

BRAIN, cilt.144, ss.1422-1434, 2021 (SCI-Expanded)

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.

FRONTIERS IN CELL AND DEVELOPMENTAL BIOLOGY, cilt.9, 2021 (SCI-Expanded)

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.

PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA, cilt.118, sa.15, 2021 (SCI-Expanded)

XXXII. **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.

BMC MEDICAL GENOMICS, cilt.14, sa.1, 2021 (SCI-Expanded)

XXXIII. **METAP1mutation is a novel candidate for autosomal recessive intellectual disability**

ÇAĞLAYAN A. O., Aktar F., Bilguvar K., Baranoski J. F., Akgumus G. T., Harmanci A. S., Erson-Omay E. Z., Yasuno K., Caksen H., Gunel M.

JOURNAL OF HUMAN GENETICS, cilt.66, sa.2, ss.215-218, 2021 (SCI-Expanded)

XXXIV. **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)

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.

JOURNAL OF EXPERIMENTAL MEDICINE, cilt.218, sa.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.
CLINICAL DYSMORPHOLOGY, cilt.30, sa.1, ss.54-57, 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. **Mutation spectrum of congenital heart disease in 73consanguineous Turkish families**
Dong W., Kaymakcalan H., Diab N., Jin S. C., Tanidir C., Yalcin A., Mane S., Bilguvar K., Brueckner M., Lifton R.
EUROPEAN JOURNAL OF HUMAN GENETICS, sa.SUPPL 1, ss.263-264, 2020 (SCI-Expanded)

XXXIX. **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.
BIOMEDICINES, cilt.8, sa.12, 2020 (SCI-Expanded)

XL. **Correlations between genomic subgroup and clinical features in a cohort of more than 3000 meningiomas**
Youngblood M. W., Duran D., Montejo J. D., Li C., Omay S. B., ÖZDUMAN K., Sheth A. H., Zhao A. Y., Tyrtova E., Miyagishima D. F., et al.
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. **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.
SCIENCE, cilt.370, sa.6515, 2020 (SCI-Expanded)

XLIII. **Autoantibodies against type I IFNs in patients with life-threatening COVID-19**
Bastard P., Rosen L. B., Zhang Q., Michailidis E., Hoffmann H., Zhang Y., Dorgham K., Philippot Q., Rosain J., Beziat V., et al.
SCIENCE, cilt.370, sa.6515, ss.423-435, 2020 (SCI-Expanded)

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.
NATURE GENETICS, cilt.52, sa.10, ss.1046-1056, 2020 (SCI-Expanded)

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.
JOURNAL OF CLINICAL INVESTIGATION, cilt.130, sa.7, ss.3885-3900, 2020 (SCI-Expanded)

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

XLVII. **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.

BRAIN, cilt.143, ss.1447-1461, 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**
Duran D., Zeng X., Jin S. C., Choi J., Nelson-Williams C., Yatsula B., Gaillard J., Furey C. G., Lu Q., Timberlake A. T., et al.
NEURON, cilt.101, sa.3, ss.429-447, 2019 (SCI-Expanded)

L. **Whole-exome sequencing of cervical carcinomas identifies activating ERBB2 and PIK3CA mutations as targets for combination therapy**
Zammataro L., Lopez S., Bellone S., Pettinella F., Bonazzoli E., Perrone E., Zhao S., Menderes G., Altwerger G., Han C., et al.
PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA, cilt.116, sa.45, ss.22730-22736, 2019 (SCI-Expanded)

LI. **Clonal evolution analysis of paired anaplastic and well-differentiated thyroid carcinomas reveals shared common ancestor**
Dong W., Nicolson N. G., Choi J., Barbieri A. L., Kunstman J. W., Abou Azar S., Knight J., Bilguvar K., Mane S. M., Lifton R. P., et al.
GENES CHROMOSOMES & CANCER, cilt.57, sa.12, ss.645-652, 2018 (SCI-Expanded)

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.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.103, sa.5, ss.666-678, 2018 (SCI-Expanded)

LIV. **Meningioma With Multiple Drivers: Genomic Landscape and Clinical Correlations**
Tyrtova E., Li C., Youngblood M., Duran D., Montejo J. D., Coskun S., Miyagishima D. F., Bilguvar K., Gunel M.
NEUROSURGERY, ss.94, 2018 (SCI-Expanded)

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.
NATURE GENETICS, cilt.50, sa.8, ss.1093-1107, 2018 (SCI-Expanded)

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.**
Harmancı 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.
Nature communications, cilt.9, ss.16215, 2018 (SCI-Expanded)

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., Bilguv K., Vural A., Basak A. N.
NEUROLOGY-GENETICS, cilt.4, 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.
NATURE GENETICS, cilt.49, sa.11, ss.1593-1601, 2017 (SCI-Expanded)

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, cilt.20, 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.
ANNALS OF NEUROLOGY, cilt.82, sa.3, ss.466-478, 2017 (SCI-Expanded)

LXIII. **Familial occurrence of brain arteriovenous malformation: a novel ACVRL1 mutation detected by whole exome sequencing**
Yilmaz B., Toktas Z. O., Akakin A., Isik S., Bilguvar K., Kilic T., Gunel M.
JOURNAL OF NEUROSURGERY, cilt.126, sa.6, ss.1879-1883, 2017 (SCI-Expanded)

LXIV. **Biallelic mutations in the 3′ exonuclease TOE1 cause pontocerebellar hypoplasia and uncover a role in snRNA processing**
Lardelli R. M., Schaffer A. E., Eggens V. R. C., Zaki M. S., Grainger S., Sathe S., Van Nostrand E. L., Schlachetzki Z., Rosti B., Akizu N., et al.
NATURE GENETICS, cilt.49, sa.3, ss.457-464, 2017 (SCI-Expanded)

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.
NATURE COMMUNICATIONS, cilt.8, 2017 (SCI-Expanded)

LXVI. **Longitudinal analysis of treatment-induced genomic alterations in gliomas**
Erson-Omay E. Z., Henegariu O., Omay S. B., Harmanci A. S., Youngblood M. W., Mishra-Gorur K., Li J., ÖZDUMAN K., Carrion-Grant G., Clark V. E., et al.
GENOME MEDICINE, cilt.9, 2017 (SCI-Expanded)

LXVII. **Impaired Amino Acid Transport at the Blood Brain Barrier Is a Cause of Autism Spectrum Disorder**
Tarlungeanu D. C., Deliu E., Dotter C. P., Kara M., Janiesch P. C., Scalise M., Galluccio M., Tesulov M., Morelli E., Sonmez F. M., et al.
CELL, cilt.167, sa.6, ss.1481-1512, 2016 (SCI-Expanded)

LXVIII. **Biallelic Mutations in TMTC3, Encoding a Transmembrane and TPR-Containing Protein, Lead to Cobblestone Lissencephaly**
Jerber J., Zaki M. S., Al-Aama J. Y., Rosti R. O., Ben-Omran T., Dikoglu E., Silhavy J. L., ÇAĞLAR C., Musaev D., Albrecht B., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.99, sa.5, ss.1181-1189, 2016 (SCI-Expanded)

LXIX. **Mutational landscape of uterine and ovarian carcinosarcomas implicates histone genes in epithelial-mesenchymal transition**
Zhao S., Bellone S., Lopez S., Thakral D., Schwab C., English D. P., Black J., Cocco E., Choi J., Zammataro L., et al.
PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA, cilt.113, sa.43, ss.12238-12243, 2016 (SCI-Expanded)

LXX. **ACOX2 deficiency: A disorder of bile acid synthesis with transaminase elevation, liver fibrosis, ataxia, and cognitive impairment**
Vilarinho S., SARI S., Mazzacuva F., Bilguvar K., Esendagli-Yilmaz G., Jain D., AKYOL G., DALGIÇ B., Gunel M., Clayton P. T., et al.

PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA, cilt.113, sa.40, ss.11289-11293, 2016 (SCI-Expanded)

LXXI. **Recurrent somatic mutations in POLR2A define a distinct subset of meningiomas**
Clarke V. E., Harmanci A. S., Bai H., Youngblood M. W., Lee T. I., Baranoski J. F., Ercan-Sencicek A. G., Abraham B. J., Weintraub A. S., Hnisz D., et al.
NATURE GENETICS, cilt.48, sa.10, ss.1253-1259, 2016 (SCI-Expanded)

LXXII. **Two locus inheritance of non-syndromic midline craniosynostosis via rare SMAD6 and common BMP2 alleles**
Timberlake A. T., Choi J., Zaidi S., Lu Q., Nelson-Williams C., Brooks E. D., Bilguvar K., Tikhonova I., Mane S., Yang J. F., et al.
ELIFE, cilt.5, 2016 (SCI-Expanded)

LXXIII. **Biallelic Mutations in Citron Kinase Link Mitotic Cytokinesis to Human Primary Microcephaly**
Li H., Bielas S. L., Zaki M. S., Ismail S., Farfara D., Um K., Rosti R. O., Scott E. C., Tu S., Chi N. C., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.99, sa.2, ss.501-510, 2016 (SCI-Expanded)

LXXIV. **Loss-of-Function Mutations in FRRS1L Lead to an Epileptic-Dyskinetic Encephalopathy**
Madeo M., Stewart M., Sun Y., Sahir N., Wiethoff S., Chandrasekar I., Yarrow A., Rosenfeld J. A., Yang Y., Cordeiro D., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.98, sa.6, ss.1249-1255, 2016 (SCI-Expanded)

LXXV. **Overlapping 16p13.11 Deletion and Gain of Copies Variations Associated with Childhood Onset Psychosis Include Genes with Mechanistic Implications for Autism Associated Pathways: Two Case Reports**
Brownstein C. A., Kleiman R. J., Engle E. C., Towne M. C., D'Angelo E. J., Yu T. W., Beggs A. H., Picker J., Fogler J. M., Carroll D., et al.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.170, sa.5, ss.1165-1173, 2016 (SCI-Expanded)

LXXVI. **A patient with a novel homozygous missense mutation in FTO and concomitant nonsense mutation in CETP**
ÇAĞLAYAN A. O., Tuysuz B., Coskun S., Quon J., Harmanci A. S., Baranoski J. F., BARAN B., Erson-Omay E. Z., Henegariu O., Mane S. M., et al.
JOURNAL OF HUMAN GENETICS, cilt.61, sa.5, ss.395-403, 2016 (SCI-Expanded)

LXXVII. **Renal Involvement in Patients with Mucolipidosis IIIAlpha/Beta: Causal Relation or Co-Occurrence?**
Tuysuz B., Ercan-Sencicek A. G., CANPOLAT N., Koparir A., Yilmaz S., KILIÇASLAN I., Gulez B., Bilguvar K., Gunel M.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.170, sa.5, ss.1187-1195, 2016 (SCI-Expanded)

LXXVIII. **Absence of KMT2D/MLL2 mutations in abdominal paraganglioma**
Bilguvar K., Goh G., Korah R., Lifton R. P., Carling T.
CLINICAL ENDOCRINOLOGY, cilt.84, sa.4, ss.632-634, 2016 (SCI-Expanded)

LXXIX. **Genomic characterization of sarcomatoid transformation in clear cell renal cell carcinoma**
Bi M., Zhao S., Said J. W., Merino M. J., Adeniran A. J., Xie Z., Nawaf C. B., Choi J., Belldegrun A. S., Pantuck A. J., et al.
PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA, cilt.113, sa.8, ss.2170-2175, 2016 (SCI-Expanded)

LXXX. **Integrated genomic characterization of IDH1-mutant glioma malignant progression**
Bai H., Harmanci A. S., Erson-Omay E. Z., Li J., Coskun S., Simon M., Krischek B., ÖZDUMAN K., Omay S. B., Sorensen E. A., et al.
NATURE GENETICS, cilt.48, sa.1, ss.59-69, 2016 (SCI-Expanded)

LXXXI. **De novo mutations in congenital heart disease with neurodevelopmental and other congenital anomalies**
Homsy J., Zaidi S., Shen Y., Ware J. S., Samocha K. E., Karczewski K. J., DePalma S. R., McKean D., Wakimoto H., Gorham J., et al.
SCIENCE, cilt.350, sa.6265, ss.1262-1266, 2015 (SCI-Expanded)

LXXXII. **Clinical, Electrodiagnostic, and Genetic Features of Tangier Disease in an Adolescent Girl with Presentation of Peripheral Neuropathy**
PER H., CANPOLAT M., Bayram A. K., Ulgen E., BARAN B., KARDAŞ F., GÜMÜŞ H., Kumandas S., Bilguvar K.,

ÇAĞLAYAN A. O.
NEUROPEDIATRICS, cilt.46, sa.6, ss.420-423, 2015 (SCI-Expanded)

LXXXIII. **Somatic V600E BRAF Mutation in Linear and Sporadic Syringocystadenoma Papilliferum**
Levinsohn J. L., Sugarman J. L., Bilguvar K., McNiff J. M., Choate K. A.
JOURNAL OF INVESTIGATIVE DERMATOLOGY, cilt.135, sa.10, ss.2536-2538, 2015 (SCI-Expanded)

LXXXIV. **Somatic POLE mutations cause an ultramutated giant cell high-grade glioma subtype with better prognosis**
Erson-Omay E. Z., Caglayan A. O., Schultz N., Weinhold N., Omay S. B., ÖZDUMAN K., Koksal Y., Li J., Harmanci A. S., Clark V., et al.
NEURO-ONCOLOGY, cilt.17, sa.10, ss.1356-1364, 2015 (SCI-Expanded)

LXXXV. **Whole-exome sequencing defines the mutational landscape of pheochromocytoma and identifies KMT2D as a recurrently mutated gene**
Juhlin C. C., Stenman A., Haglund F., Clark V. E., Brown T. C., Baranoski J., Bilguvar K., Goh G., Welander J., Svahn F., et al.
GENES CHROMOSOMES & CANCER, cilt.54, sa.9, ss.542-554, 2015 (SCI-Expanded)

LXXXVI. **The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities**
Chong J. X., Buckingham K. J., Jhangiani S. N., Boehm C., Sobreira N., Smith J. D., Harrell T. M., McMillin M. J., Wiszniewski W., Gambin T., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.97, sa.2, ss.199-215, 2015 (SCI-Expanded)

LXXXVII. **Exome sequencing links mutations in PARN and RTEL1 with familial pulmonary fibrosis and telomere shortening**
Stuart B. D., Choi J., Zaidi S., Xing C., Holohan B., Chen R., Choi M., Dharwadkar P., Torres F., Girod C. E., et al.
NATURE GENETICS, cilt.47, sa.5, ss.512-517, 2015 (SCI-Expanded)

LXXXVIII. **Spondyloepimetaphyseal dysplasia with joint laxity, leptodactylic type: longitudinal observation of radiographic findings in a child heterozygous for a KIF22 mutation**
Tuysuz B., Yilmaz S., Erener-Ercan T., Bilguvar K., Gunel M.
PEDIATRIC RADIOLOGY, cilt.45, sa.5, ss.771-776, 2015 (SCI-Expanded)

LXXXIX. **Functional Synergy between Cholecystokinin Receptors CCKAR and CCKBR in Mammalian Brain Development**
Nishimura S., Bilguevar K., Ishigame K., Sestan N., Guenel M., Louvi A.
PLOS ONE, cilt.10, sa.4, 2015 (SCI-Expanded)

XC. **The distinct genetic pattern of ALS in Turkey and novel mutations**
Ozoguz A., Uyan O., Birdal G., Iskender C., Kartal E., Lahut S., Omur O., Agim Z. S., Eken A. G., Sen N. E., et al.
NEUROBIOLOGY OF AGING, cilt.36, sa.4, 2015 (SCI-Expanded)

XCI. **Vascular Endothelial Growth Factor Receptor 3 Controls Neural Stem Cell Activation in Mice and Humans**
Han J., Calvo C., Kang T. H., Baker K. L., Park J., Parras C., Levittas M., Birba U., Pibouin-Fragner L., Fragner P., et al.
CELL REPORTS, cilt.10, sa.7, ss.1158-1172, 2015 (SCI-Expanded)

XCII. **Homozygous loss of DIAPH1 is a novel cause of microcephaly in humans**
Ercan-Sencicek A. G., Jambi S., Franjic D., Nishimura S., Li M., El-Fishawy P., Morgan T. M., Sanders S. J., Bilguvar K., Suri M., et al.
EUROPEAN JOURNAL OF HUMAN GENETICS, cilt.23, sa.2, ss.165-172, 2015 (SCI-Expanded)

XCIII. **Dominant De Novo Mutations in GJA1 Cause Erythrokeratodermia Variabilis et Progressiva, without Features of Oculodentodigital Dysplasia**
Boyden L. M., Craiglow B. G., Zhou J., Hu R., Loring E. C., Morel K. D., Lauren C. T., Lifton R. P., Bilguvar K., Paller A. S., et al.
JOURNAL OF INVESTIGATIVE DERMATOLOGY, cilt.135, sa.6, ss.1540-1547, 2015 (SCI-Expanded)

XCIV. **A rare case of congenital fibrosis of extraocular muscle type IA due to KIF21A mutation with Marcus Gunn jaw-winking phenomenon**
Bayram A. K., PER H., Quon J., CANPOLAT M., Uelgen E., Dogan H., GÜMÜŞ H., Kumandas S., Bayram N., Bilguvar K., et al.

EUROPEAN JOURNAL OF PAEDIATRIC NEUROLOGY, cilt.19, sa.6, ss.743-746, 2015 (SCI-Expanded)

XCV. **NGLY1 mutation causes neuromotor impairment, intellectual disability, and neuropathy**
ÇAĞLAYAN A. O., Comu S., Baranoski J. F., Parman Y., Kaymakcalan H., Akgumus G. T., ÇAĞLAR C., Dolen D., Erson-Omay E. Z., Harmanci A. S., et al.
EUROPEAN JOURNAL OF MEDICAL GENETICS, cilt.58, sa.1, ss.39-43, 2015 (SCI-Expanded)

XCVI. **Mutations in KATNB1 Cause Complex Cerebral Malformations by Disrupting Asymmetrically Dividing Neural Progenitors**
Mishra-Gorur K., ÇAĞLAYAN A. O., Schaffer A. E., Chabu C., Henegariu O., Vonhoff F., Akguemues G. T., Nishimura S., Han W., Tu S., et al.
NEURON, cilt.84, sa.6, ss.1226-1239, 2014 (SCI-Expanded)

XCVII. **Brain Malformations Associated With Knobloch Syndrome-Review of Literature, Expanding Clinical Spectrum, and Identification of Novel Mutations**
ÇAĞLAYAN A. O., Baranoski J. E., Aktar F., Han W., Tuysuz B., Guzel A., Guclu B., Kaymakcalan H., Aktekin B., Akgumus G. T., et al.
PEDIATRIC NEUROLOGY, cilt.51, sa.6, ss.806-813, 2014 (SCI-Expanded)

XCVIII. **FBX07-R498X mutation: Phenotypic variability from chorea to early onset parkinsonism within a family**
GÜNDÜZ A., Eken A. G., BİLGİÇ B., HANAĞASI H. A., Bilguvar K., Guenel M., Basak A. N., Ertan S.
PARKINSONISM & RELATED DISORDERS, cilt.20, sa.11, ss.1253-1256, 2014 (SCI-Expanded)

XCIX. **Primary hypertrophic osteoarthropathy caused by homozygous deletion in HPGD gene in a family: changing clinical and radiological findings with long-term follow-up**
Tuysuz B., Yilmaz S., KASAPÇOPUR Ö., Erener-Ercan T., Ceyhun E., Bilguvar K., Gunel M.
RHEUMATOLOGY INTERNATIONAL, cilt.34, sa.11, ss.1539-1544, 2014 (SCI-Expanded)

C. **CLP1 Founder Mutation Links tRNA Splicing and Maturation to Cerebellar Development and Neurodegeneration**
Schaffer A. E., Eggens V. R. C., ÇAĞLAYAN A. O., Reuter M. S., Scott E., Coufal N. G., Silhavy J. L., Xue Y., Kayserili H., Yasuno K., et al.
CELL, cilt.157, sa.3, ss.651-663, 2014 (SCI-Expanded)

CI. **Spontaneous tumour regression in keratoacanthomas is driven by Wnt/retinoic acid signalling cross-talk**
Zito G., Saotome I., Liu Z., Ferro E. G., Sun T. Y., Nguyen D. X., Bilguvar K., Ko C. J., Greco V.
NATURE COMMUNICATIONS, cilt.5, 2014 (SCI-Expanded)

CII. **Paediatric hepatocellular carcinoma due to somatic CTNNB1 and NFE2L2 mutations in the setting of inherited bi-allelic ABCB11 mutations**
Vilarinho S., Erson-Omay E. Z., Harmanci A. S., Morotti R., Carrion-Grant G., Baranoski J., Knisely A. S., Ekong U., Emre S., Yasuno K., et al.
JOURNAL OF HEPATOLOGY, cilt.61, sa.5, ss.1178-1183, 2014 (SCI-Expanded)

CIII. **Exome Sequencing Links Corticospinal Motor Neuron Disease to Common Neurodegenerative Disorders**
Novarino G., Fenstermaker A. G., Zaki M. S., Hofree M., Silhavy J. L., Heiberg A. D., Abdellateef M., Rosti B., Scott E., Mansour L., et al.
SCIENCE, cilt.343, sa.6170, ss.506-511, 2014 (SCI-Expanded)

CIV. **Autosomal recessive spastic tetraplegia caused by AP4M1 and AP4B1 gene mutation: Expansion of the facial and neuroimaging features**
Tuysuz B., Bilguvar K., Kocer N., YALÇINKAYA C., ÇAĞLAYAN A. O., Gul E., ŞAHİN S., Comu S., Gunel M.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.164, sa.7, ss.1677-1685, 2014 (SCI-Expanded)

CV. **A new patient with andermann syndrome: An underdiagnosed clinical genetics entity?**
Degerliyurt A., Akgumus G., Caglar C., Bilguvar K., Caglayan A.
Genetic Counseling, cilt.24, sa.3, ss.283-289, 2013 (SCI-Expanded)

CVI. **Whole-exome sequencing identified a patient with TMCO1 defect syndrome and expands the phenotic spectrum**

ÇAĞLAYAN A. O., PER H., Akgumus G., GÜMÜŞ H., Baranoski J., CANPOLAT M., Calik M., Yikilmaz A., Bilguvar K., Kumandas S., et al.
CLINICAL GENETICS, cilt.84, sa.4, ss.394-395, 2013 (SCI-Expanded)

CVII. **FBXO7 mutation: Phenotypic variability from chorea to early-onset asymmetric parkinsonism within a family**
GÜNDÜZ A., Eken A. G., Bilguvar K., KEREM GÜNEL M., Basak A. N., HANAĞASI H. A., Ertan S.
MOVEMENT DISORDERS, 2013 (SCI-Expanded)

CVIII. **Mutations in LAMB1 Cause Cobblestone Brain Malformation without Muscular or Ocular Abnormalities**
Radmanesh F., ÇAĞLAYAN A. O., Silhavy J. L., Yilmaz C., Cantagrel V., Omar T., Rosti B., Kaymakcalan H., Gabriel S., Li M., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.92, sa.3, ss.468-474, 2013 (SCI-Expanded)

CIX. **Missense mutation in the ATPase, aminophospholipid transporter protein ATP8A2 is associated with cerebellar atrophy and quadrupedal locomotion**
Onat O. E., Gulsuner S., Bilguvar K., Basak A. N., Topaloglu H., Tan M., Tan U., Gunel M., Ozcelik T.
EUROPEAN JOURNAL OF HUMAN GENETICS, cilt.21, sa.3, ss.281-285, 2013 (SCI-Expanded)

CX. **Genomic Analysis of Non-NF2 Meningiomas Reveals Mutations in TRAF7, KLF4, AKT1, and SMO**
Clark V. E., Erson-Omay E. Z., Serin A., Yin J., Cotney J., ÖZDUMAN K., Avsar T., Li J., Murray P. B., Henegariu O., et al.
SCIENCE, cilt.339, sa.6123, ss.1077-1080, 2013 (SCI-Expanded)

CXI. **Recessive loss of function of the neuronal ubiquitin hydrolase UCHL1 leads to early-onset progressive neurodegeneration**
Bilguvar K., Tyagi N. K., ÖZKARA Ç., Tuysuz B., Bakircioglu M., Choi M., Delil S., ÇAĞLAYAN A. O., Baranoski J. F., Erturk O., et al.
PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA, cilt.110, sa.9, ss.3489-3494, 2013 (SCI-Expanded)

CXII. **Spondyloepimetaphyseal Dysplasia Pakistani Type: Expansion of the Phenotype**
Tuysuz B., Yilmaz S., Gul E., Kolb L., Bilguvar K., EVLİYAOĞLU S. O., Gunel M.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.161, sa.6, ss.1300-1308, 2013 (SCI-Expanded)

CXIII. **De novo mutations revealed by whole-exome sequencing are strongly associated with autism**
Sanders S. J., Murtha M. T., Gupta A. R., Murdoch J. D., Raubeson M. J., Willsey A. J., Ercan-Sencicek A. G., DiLullo N. M., Parikshak N. N., Stein J. L., et al.
NATURE, cilt.485, sa.7397, ss.237-241, 2012 (SCI-Expanded)

CXIV. **Rare Copy Number Variants in Tourette Syndrome Disrupt Genes in Histaminergic Pathways and Overlap with Autism**
Fernandez T. V., Sanders S. J., Yurkiewicz I. R., Ercan-Sencicek A. G., Kim Y., Fishman D. O., Raubeson M. J., Song Y., Yasuno K., Ho W. S. C., et al.
BIOLOGICAL PSYCHIATRY, cilt.71, sa.5, ss.392-402, 2012 (SCI-Expanded)

CXV. **Common variant near the endothelin receptor type A (EDNRA) gene is associated with intracranial aneurysm risk**
Yasuno K., Bakircioglu M., Low S., Bilguevar K., Gaal E., Ruigrok Y. M., Niemela M., Hata A., Bijlenga P., Kasuya H., et al.
PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA, cilt.108, sa.49, ss.19707-19712, 2011 (SCI-Expanded)

CXVI. **Homozygosity mapping and targeted genomic sequencing reveal the gene responsible for cerebellar hypoplasia and quadrupedal locomotion in a consanguineous kindred**
Gulsuner S., Tekinay A. B., Doerschner K., Boyaci H., Bilguvar K., Unal H., Ors A., Onat O. E., Atalar E., Basak A. N., et al.
GENOME RESEARCH, cilt.21, sa.12, ss.1995-2003, 2011 (SCI-Expanded)

CXVII. **Multiple Recurrent De Novo CNVs, Including Duplications of the 7q11.23 Williams Syndrome Region, Are Strongly Associated with Autism**

Sanders S. J., Ercan-Sencicek A. G., Hus V., Luo R., Murtha M. T., Moreno-De-Luca D., Chu S. H., Moreau M. P., Gupta A. R., Thomson S. A., et al.
NEURON, cilt.70, sa.5, ss.863-885, 2011 (SCI-Expanded)

CXVIII. **Recessive LAMC3 mutations cause malformations of occipital cortical development**
Barak T., Kwan K. Y., Louvi A., Demirbilek V., SAYGI S., Tuysuz B., Choi M., Boyaci H., Doerschner K., Zhu Y., et al.
NATURE GENETICS, cilt.43, sa.6, ss.590-596, 2011 (SCI-Expanded)

CXIX. **The Essential Role of Centrosomal NDE1 in Human Cerebral Cortex Neurogenesis**
Bakircioglu M., Carvalho O. P., Khurshid M., Cox J. J., Tuysuz B., Barak T., Yilmaz S., ÇAĞLAYAN A. O., DİNÇER A., Nicholas A. K., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.88, sa.5, ss.523-535, 2011 (SCI-Expanded)

CXX. **Four Novel SCN1A Mutations in Turkish Patients With Severe Myoclonic Epilepsy of Infancy (SMEI)**
Arlier Z., Bayri Y., Kolb L. E., Erturk O., Ozturk A. K., Bayrakli F., Bilguvar K., Moliterno J. A., Dervent A., Demirbilek V., et al.
JOURNAL OF CHILD NEUROLOGY, cilt.25, sa.10, ss.1265-1268, 2010 (SCI-Expanded)

CXXI. **Whole-exome sequencing identifies recessive WDR62 mutations in severe brain malformations**
Bilguvar K., Ozturk A. K., Louvi A., Kwan K. Y., Choi M., Tatli B., YALNIZOĞLU D., Tuysuz B., ÇAĞLAYAN A. O., Gokben S., et al.
NATURE, cilt.467, sa.7312, ss.207-211, 2010 (SCI-Expanded)

CXXII. **Novel VLDLR microdeletion identified in two Turkish siblings with pachygyria and pontocerebellar atrophy**
Kolb L. E., Arlier Z., YALÇINKAYA C., Ozturk A. K., Moliterno J. A., Erturk O., Bayrakli F., KORKMAZ M. B., DiLuna M. L., Yasuno K., et al.
NEUROGENETICS, cilt.11, sa.3, ss.319-325, 2010 (SCI-Expanded)

CXXIII. **L-Histidine Decarboxylase and Tourette's Syndrome**
Ercan-Sencicek A. G., Stillman A. A., Ghosh A. K., Bilguvar K., O'Roak B. J., Mason C. E., Abbott T., Gupta A., King R. A., Pauls D. L., et al.
NEW ENGLAND JOURNAL OF MEDICINE, cilt.362, sa.20, ss.1901-1908, 2010 (SCI-Expanded)

CXXIV. **Genome-wide association study of intracranial aneurysm identifies three new risk loci**
Yasuno K., Bilguvar K., Bijlenga P., Low S., Krischek B., Auburger G., Simon M., Krex D., Arlier Z., Nayak N., et al.
NATURE GENETICS, cilt.42, sa.5, ss.420-425, 2010 (SCI-Expanded)

CXXV. **Heterozygous 5p13.3-13.2 deletion in a patient with type I Chiari malformation and bilateral Duane retraction syndrome**
Bayrakli F., Bilguvar K., Ceyhan D., Ercan-Sencicek A. G., ÇANKAYA T., Bayrakli S., GÜNEY A. İ., Mane S. M., State M. W., Gunel M.
CLINICAL GENETICS, cilt.77, sa.5, ss.499-502, 2010 (SCI-Expanded)

CXXVI. **A Patient With Duchenne Muscular Dystrophy and Autism Demonstrates a Hemizygous Deletion Affecting Dystrophin**
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.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.152, sa.4, ss.1039-1042, 2010 (SCI-Expanded)

CXXVII. **A novel heterozygous deletion within the 3′ region of the PAX6 gene causing isolated aniridia in a large family group**
Bayrakli F., GÜNEY A. İ., Bayri Y., Ercan-Sencicek A. G., Ceyhan D., ÇANKAYA T., Mason C., Bilguvar K., Bayrakli S., Mane S. M., et al.
JOURNAL OF CLINICAL NEUROSCIENCE, cilt.16, sa.12, ss.1610-1614, 2009 (SCI-Expanded)

CXXVIII. **The Syndrome of Pachygyria, Mental Retardation, and Arachnoid Cysts Maps to 11p15**
Bilguvar K., Ozturk A. K., Bayrakli F., Guzel A., DiLuna M. L., Bayri Y., Tatli M., Tekes S., Arlier Z., Yasuno K., et al.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.149, sa.11, ss.2569-2572, 2009 (SCI-Expanded)

CXXIX. **COL4A1 Mutation in Preterm Intraventricular Hemorrhage**
Bilguvar K., DiLuna M. L., Bizzarro M. J., Bayri Y., Schneider K. C., Lifton R. P., Gunel M., Ment L. R.

JOURNAL OF PEDIATRICS, cilt.155, sa.5, ss.743-745, 2009 (SCI-Expanded)

CXXX. **Susceptibility loci for intracranial aneurysm in European and Japanese populations**
Bilguvar K., Yasuno K., Niemela M., Ruigrok Y. M., Fraunberg M. v. u. z., van Duijn C. M., van den Berg L. H., Mane S., Mason C. E., Choi M., et al.
NATURE GENETICS, cilt.40, sa.12, ss.1472-1477, 2008 (SCI-Expanded)

CXXXI. **Therapeutic efficacy of SJA6017, a calpain inhibitor, in rat spinal cord injury**
Akdemir O., Ucankale M., Karaoglan A., Barut S., Sagmanligil A., Bilguvar K., Cirakoglu B., Sahan E., Colak A.
JOURNAL OF CLINICAL NEUROSCIENCE, cilt.15, sa.10, ss.1130-1136, 2008 (SCI-Expanded)

CXXXII. **Neuroprotective effects of Ac.YVAD.cmk on experimental spinal cord injury in rats**
Karaoglan A., KAYA E., Akdemir O., Sagmanligil A., Bilguvar K., Cirakoglu B., Sahan E., Erdogan N., Barut S., Colak A.
SURGICAL NEUROLOGY, cilt.69, sa.6, ss.561-567, 2008 (SCI-Expanded)

CXXXIII. **Therapeutic efficacy of Ac-DMQD-CHO, a caspase 3 inhibitor, for rat spinal cord injury**
Akdemir O., Berksoy I., Karaoglan A., Barut S., Bilguvar K., Cirakoglu B., Sahan E., Colak A.
JOURNAL OF CLINICAL NEUROSCIENCE, cilt.15, sa.6, ss.672-678, 2008 (SCI-Expanded)

CXXXIV. **Novel NTRK1 mutations cause hereditary sensory and autonomic neuropathy type IV:: demonstration of a founder mutation in the Turkish population**
Tuysuz B., Bayrakli F., DiLuna M. L., Bilguvar K., Bayri Y., YALÇINKAYA C., Bursali A., Ozdamar E., KORKMAZ M. B., Mason C. E., et al.
NEUROGENETICS, cilt.9, sa.2, ss.119-125, 2008 (SCI-Expanded)

CXXXV. **A novel syndrome of cerebral cavernous malformation and Greig cephalopolysyndactyly**
Bilguvar K., Bydon M., Bayrakli F., Ercan-Sencicek A. G., Bayri Y., Mason C., Diluna M. L., Seashore M., Bronen R., Lifton R. P., et al.
JOURNAL OF NEUROSURGERY, cilt.107, sa.6, ss.495-499, 2007 (SCI-Expanded)

CXXXVI. **Rapid identification of disease-causing mutations using copy number analysis within linkage intervals**
Bayrakli F., Bilguvar K., Mason C. E., DiLuna M. L., Bayri Y., Gungor L., Terzi M., Mane S. M., Lifton R. P., State M. W., et al.
HUMAN MUTATION, cilt.28, sa.12, ss.1236-1240, 2007 (SCI-Expanded)

CXXXVII. **Apparently novel genetic syndrome of pachygyria, mental retardation, seizure, and arachnoid cysts**
Guzel A., Tatli M., Bilguvar K., DiLuna M. L., Bakkaloglu B., Ozturk A. K., Bayrakli F., Gunel M.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.143A, sa.7, ss.672-677, 2007 (SCI-Expanded)

CXXXVIII. **Genetics of intracranial aneurysms**
Nahed B. V., Bydon M., Ozturk A. K., Bilguvar K., Bayrakli F., Gunel M.
NEUROSURGERY, cilt.60, sa.2, ss.213-225, 2007 (SCI-Expanded)

CXXXIX. **Controversial molecular classification of human cerebrovascular malformations - Response to letter by Stahl and Felbor**
Bilguvar K., Ozturk A. K., Gunel M., Guclu B.
STROKE, cilt.37, sa.9, ss.2215-2216, 2006 (SCI-Expanded)

CXL. **Molecular genetic analysis of two large kindreds with intracranial aneurysms demonstrates linkage to 11q24-25 and 14q23-31**
Ozturk A., Nahed B., Bydon M., Bilguvar K., Goksu E., Bademci G., Guclu B., Johnson M., Amar A., Lifton R., et al.
STROKE, cilt.37, sa.4, ss.1021-1027, 2006 (SCI-Expanded)

CXLI. **Mutations in apoptosis-related gene, PDCD10, cause cerebral cavernous malformation 3**
Guclu B., Ozturk A., Pricola K., Bilguvar K., Shin D., O'Roak B., Gunel M.
NEUROSURGERY, cilt.57, sa.5, ss.1008-1012, 2005 (SCI-Expanded)

Diğer Dergilerde Yayınlanan Makaleler

I. **Severe Phenotype in Patients with X-linked Hydrocephalus Caused by a Missense Mutation in L1CAM**

Tuysuz B., Ercan-Sencicek A. G., Ozer E., Goc N., Yalcinkaya C., Bilguvar K.
TURKISH ARCHIVES OF PEDIATRICS, cilt.57, sa.5, ss.521-525, 2022 (ESCI)

II. **ALPK3 gene mutation in a patient with congenital cardiomyopathy and dysmorphic features**
ÇAĞLAYAN A. O., Sezer R. G., Kaymakcalan H., Ulgen E., Yavuz T., Baranoski J. F., Bozaykut A., Harmanci A. S., Yalcin Y., Youngblood M. W., et al.
COLD SPRING HARBOR MOLECULAR CASE STUDIES, cilt.3, sa.5, 2017 (ESCI)

III. **Digenic mutations of human OCRL paralogs in Dent's disease type 2 associated with Chiari i malformation**
Duran D., Jin S. C., Despenza T., Nelson-Williams C., Cogal A. G., Abrash E. W., Harris P. C., Lieske J. C., Shimshak S. J., Mane S., et al.
Human Genome Variation, cilt.3, 2016 (Scopus)

IV. **A novel de novo mutation in ATP1A3 and childhood-onset schizophrenia**
Smedemark-Margulies N., Brownstein C. A., Vargas S., Tembulkar S. K., Towne M. C., Shi J., Gonzalez-Cuevas E., Liu K. X., Bilguvar K., Kleiman R. J., et al.
COLD SPRING HARBOR MOLECULAR CASE STUDIES, sa.5, 2016 (ESCI)

V. **Mutation in GM2A Leads to a Progressive Chorea-dementia Syndrome**
Salih M. A., Seidahmed M. Z., El Khashab H. Y., Hamad M. H. A., Bosley T. M., Burn S., Myers A., Landsverk M. L., Crotwell P. L., Bilguvar K., et al.
TREMOR AND OTHER HYPERKINETIC MOVEMENTS, 2015 (ESCI)

Hakemli Kongre / Sempozyum Bildiri Kitaplarında Yer Alan Yayınlar

I. **Human Cerebral Organoids with PIDD1 Mutations Implicate AKT-mTOR Pathway Hypoactivity in Lissencephaly**
Zhang C., Liang D., Lam T., Narayanan A., Gunel M., Bilguvar K., Louvi A.
148th Annual Meeting American-Neurological-Association (ANA), Pennsylvania, Amerika Birleşik Devletleri, 9 - 12 Eylül 2023

II. **COMPARATIVE PHYLODYNAMICS OF SARS-COV-2 VARIANTS IN THE NORTHEAST UNITED STATES**
Petrone M., Breban M., Ott I., Alpert T., Earnest R., Watkins A., Rothman J., Kalinich C., Brito A., Murphy S., et al.
Annual Meeting of the American-Society-of-Tropical-Medicine-and-Hygiene (ASTMH), ELECTR NETWORK, 17 - 21 Kasım 2021, ss.151-152

III. **ASSOCIATIONS OF GENOMIC SUBGROUP WITH RECURRENCE IN LOW-GRADE MENINGIOMAS**
Youngblood M., Miyagishima D., Jin L., Gupte T., Li C., Duran D., Montejo J., Zhao A., Sheth A., Tyrtova E., et al.
25th Virtual Annual Scientific Meeting and Education Day of the Society-for-Neuro-Oncology (SNO), ELECTR NETWORK, 19 - 21 Kasım 2020, ss.173

IV. **Mutations and copy number alterations in diffuse gliomas are shaped by different mechanisms**
ÖZDUMAN K., Ulgen E., Karacan S., Gerlevik U., CAN Ö., Bilguvar K., Akyerli C., Yuksel Ş., Ersen-Danyeli A., Tihan T., et al.
32nd EORTC-NCI-AACR Symposium on Molecular Targets and Cancer Therapeutics, ELECTR NETWORK, 24 - 25 Ekim 2020

V. **IDH-Mutant Gliomas Differ in Distribution of Mitochondrial Genomic Alterations**
Ozduman K., Yuksel Ş., Akyerli C., Bilguvar K., Ersen-Danyeli A., Yilmaz E., Pamir M. N.
Annual Scientific Meeting of the American-Association-of-Neurological-Surgeons (AANS), Massachusetts, Amerika Birleşik Devletleri, 25 - 29 Nisan 2020, ss.84

VI. **MENINGIOMA WITH MULTIPLE DRIVERS: GENOMIC LANDSCAPE AND CLINICAL CORRELATIONS**
Li C., Tyrtova E., Youngblood M., Miyagishima D., Duran D., Montejo J., ÖZDUMAN K., Sheth A., Zhao A., Fomchenko E., et al.
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, Arizona, Amerika Birleşik Devletleri, 20 - 24 Kasım 2019, ss.141

VII. **Exome Sequencing Defines the Molecular Pathogenesis of Vein of Galen Malformation**
Kundishora A., Zeng X., Duran D., Allocco A. A., Choi J., Jin S. C., Conine S. B., Nelson-Williams C., Gaillard J., Furey C. G., et al.
Annual Meeting of the Congress-of-Neurological-Surgeons, San-Francisco, Kostarika, 19 - 23 Ekim 2019, ss.85

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**
Barak T., Sencicek A. G. E., Miyagishima D. F., Henegariu O., Gorur K. M., Bilguvar K., Gunel M.
Annual Meeting of the Congress-of-Neurological-Surgeons, San-Francisco, Kostarika, 19 - 23 Ekim 2019, ss.71

IX. **Severe speech delay in Cohen Syndrome: three novel mutations and the long-term follow-up of nine patients**
Akdeniz B., GÜNEŞ N., ULUDAĞ ALKAYA D., Ercan-Sencicek G., ÇAĞLAYAN A. O., Bilguvar K., Tuysuz B.
51st Conference of the European-Society-of-Human-Genetics (ESHG) in conjunction with the European Meeting on Psychosocial Aspects of Genetics (EMPAG), Milan, İtalya, 16 - 19 Haziran 2018, ss.315-316

X. **A DEEP SEQUENCING APPROACH TO DEFINE BENZIMIDAZOLE RESISTANCE GENE FREQUENCIES IN HUMAN HOOKWORM EGG SAMPLES FROM KPANDAI DISTRICT, GHANA**
George S., Suwondo P., Otchere J., Harrison L. M., Bilguvar K., Knight J., Caccone A., Humphries D., Wilson M. D., Cappello M.
68th Annual Meeting of the American-Society-for-Tropical-Medicine-and-Hygiene (ASTMH), Maryland, Amerika Birleşik Devletleri, 20 - 24 Kasım 2019, ss.616

XI. **Damaging Genomic Variants Constitute a Major Risk Factor for Cerebral Palsy**
Kruer M., Jin S., Bakhtiari S., Lewis S., Zeng X., Sierant M., Corbett M., Norton B., Xing X., Bilguvar K., et al.
47th Annual Meeting of the Child-Neurology-Society (CNS), Illinois, Amerika Birleşik Devletleri, 15 - 18 Ekim 2018

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.
107th Annual Meeting of the United-States-and-Canadian-Academy-of-Pathology (USCAP), Vancouver, Kanada, 17 - 23 Mart 2018, ss.399-400

XIII. **SIGNIFICANTLY MUTATED GENES FOR RADIATION-ASSOCIATED MENINGIOMA**
Claus E. B., Greenhalgh S., Gaffney S. G., Bilguvar K., Calvocoressi L., Lu L., Al-Mefty O., Zhao Z. Z., Townsend J. P.
5th Quadrennial Meeting of the World-Federation-of-Neuro-Oncology-Societies (WFNOS), Zürich, İsviçre, 4 - 07 Mayıs 2017, ss.96

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.
Annual Scientific Meeting of the American-Association-of-Neurological-Surgeons (AANS), Los-Angeles, Şili, 22 - 26 Nisan 2017

XV. **Constitutive mismatch repair defect syndrome: New insights from whole exome sequencing data and functional studies**
ÇAĞLAYAN A. O., Omay Z. E. E., Koksal Y., Coskun S., Unal E., PER H., Bilguvar K., Yasuno K., Ostergaard J. R., Gunel M.
European Biotechnology Conference, Letonya, 5 - 07 Mayıs 2016

XVI. **Familial Occurrence of Brain Arteriovenous Malformation: A Novel ACVRL1 Mutation Detected by Whole Exome Sequencing**
Toktas Z., Eksi M. S., Yilmaz B., Konya D., Bilguvar K., Guenel M., Kilic T.
83rd Annual Scientific Meeting of the American-Association-of-Neurological-Surgeons, San-Francisco, Kostarika, 5 - 09 Nisan 2014

XVII. **Somatic V600E BRAF mutation causes syringocystadenoma papilliferum**
Levinsohn J., Sugarman J., Bilguvar K., McNiff J., Choate K. A.
Annual Meeting of the Society-for-Investigative-Dermatology, Georgia, Amerika Birleşik Devletleri, 6 - 09 Mayıs 2015

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.

Joint Congress of European Neurology, İstanbul, Türkiye, 31 Mayıs - 03 Haziran 2014, ss.63

XIX. **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.
Joint Congress of European Neurology, İstanbul, Türkiye, 31 Mayıs - 03 Haziran 2014

XX. **CORRELATION OF GENETIC SIGNATURES AND HISTOLOGICAL SUBTYPES OF MENINGIOMAS**
Omay S. B., Guenel J. M., Clark V. E., Li J., Omay E. Z. E., Serin A., Kolb L. E., Hebert R. M., Bilguevar K., ÖZDUMAN K., et al.
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, Kostarika, 21 - 24 Kasım 2013, ss.160-161

XXI. **GENOMIC ANALYSIS OF TRAF7-ONLY MUTANT MENINGIOMAS REVEALS NOVEL DRIVER MUTATIONS**
Clark V., Omay Z. E., Serin A., Guenel J., Omay B., Grady C., Youngblood M., Bilguevar K., Baehring J., Piepmeier J., et al.
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, Kostarika, 21 - 24 Kasım 2013, ss.138-139

XXII. **Identification of a novel missense mutation in RAD51 in a large family with congenital mirror movements**
ONAT O. E., GÜLSÜNER S. İ., BİLGEN 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.
Annual Meeting of the Congress-of-Neuroogical-Surgeons, Louisiana, Amerika Birleşik Devletleri, 24 - 29 Ekim 2009, ss.419

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.
33rd International Stroke Conference, Louisiana, Amerika Birleşik Devletleri, 19 - 21 Şubat 2008, ss.564

XXVI. **Linkage and copy number variation analysis of large families and sibling pairs demonstrates locus heterogeneity for familial intracranial aneurysms**
Bilguvar K., Bayrakli F., Bayri Y., Ozturk A. K., DiLuna M. L., Bydon M., Gunel M.
57th Annual Congress of Neurological Surgery (CNS 2007), California, Amerika Birleşik Devletleri, 17 - 19 Eylül 2007, ss.198

XXVII. **Genetic heterogeneity of intracranial aneurysm**
Gunel M., Ozturk A., Bydon M., Nahed B., Guclu B., Bilguvar K., Goksu E., Bademci G., Amar A.
Joint Annual Meeting of the AANS/CNS Cerebrovascular Section/American-Society-of-Interventional-and-Therapeutic-Neuroradiology, Florida, Amerika Birleşik Devletleri, 17 - 20 Şubat 2006, ss.399

Metrikler

Yayın: 174
Atıf (WoS): 14884
Atıf (Scopus): 16507

H-İndeks (WoS): 52
H-İndeks (Scopus): 55