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ScopusID: 9037414300

Education Information

Doctorate, Marmara University, Institute Of Health Sciences, Turkey 2002 - 2022

Undergraduate, Marmara University, School Of Medicine, Turkey 1994 - 2000

Foreign Languages

English, C2 Mastery

Turkish, C2 Mastery

German, A1 Beginner

Research Areas

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

Academic Titles / Tasks

Assistant Professor, Acibadem Mehmet Ali Aydinlar University, School Of Medicine, Department Of Medical Sciences, 2023 - Continues

Associate Professor, Yale University, School of Medicine, Neurosurgery, 2021 - Continues

Associate Professor, Yale University, School of Medicine, Genetics, 2021 - Continues

Expert PhD, Acibadem Mehmet Ali Aydinlar University, Additional Departments, 2021 - 2024

Associate Professor, Yale University, School of Medicine, Neurosurgery, 2020 - 2021

Associate Professor, Yale University, School of Medicine, Genetics, 2019 - 2021

Assistant Professor, Yale University, School of Medicine, Genetics, 2013 - 2019

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

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

Published journal articles indexed by SCI, SSCI, and AHCI

- 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, vol.121, no.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.
BRAIN, vol.147, no.1, pp.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., Montejó J. D., Miyagishima D. F., Barak T., Nishimura S., et al.
NATURE COMMUNICATIONS, vol.14, no.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., ARGÄ K. Y., Bayrakli F.
OMICS-A JOURNAL OF INTEGRATIVE BIOLOGY, vol.27, no.9, pp.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, vol.60, no.8, pp.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, vol.120, no.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, vol.379, no.6632, pp.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, vol.12, 2023 (SCI-Expanded)
- 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, vol.5, no.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.
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- XI. **Biallelic *BICD2* variant is a novel candidate for Cohen-like syndrome**
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JOURNAL OF HUMAN GENETICS, vol.67, no.9, pp.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, vol.10, no.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, vol.119, no.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.
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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.
CELL REPORTS MEDICINE, vol.3, no.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, vol.188, no.1, pp.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, vol.27, no.12, pp.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.
GENETICS IN MEDICINE, vol.23, no.12, pp.2455-2460, 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, vol.108, no.10, pp.2006-2016, 2021 (SCI-Expanded)
- 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.
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- XXIII. **X-linked recessive TLR7 deficiency in ~1% of men under 60 years old with life-threatening COVID-19**
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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. **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, vol.185, no.7, pp.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.
JOURNAL OF INHERITED METABOLIC DISEASE, no.4, pp.1001-1012, 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.
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- XXVIII. **Associations of meningioma molecular subgroup and tumor recurrence.**
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Neuro-oncology, vol.23, no.5, pp.783-794, 2021 (SCI-Expanded)
- XXIX. **Biallelic variants in *HPDL* cause pure and complicated hereditary spastic paraplegia**
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- XXX. **Genetic Defects in *DNAH2* Underlie Male Infertility With Multiple Morphological Abnormalities of the Sperm Flagella in Humans and Mice**
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.
PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA, vol.118, no.15, 2021 (SCI-Expanded)
- XXXII. ***METAP1* mutation 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.
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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.
BMC MEDICAL GENOMICS, vol.14, no.1, 2021 (SCI-Expanded)
- 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.
CLINICAL DYSMORPHOLOGY, vol.30, no.1, pp.54-57, 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, vol.218, no.3, 2021 (SCI-Expanded)

- 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, vol.185, no.1, pp.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, pp.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.
BIOMEDICINES, vol.8, no.12, 2020 (SCI-Expanded)
- 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.
EUROPEAN JOURNAL OF HUMAN GENETICS, no.SUPPL 1, pp.263-264, 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, vol.133, no.5, pp.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, vol.26, no.11, pp.1754-1765, 2020 (SCI-Expanded)
- XLII. **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.
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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.
JOURNAL OF CLINICAL INVESTIGATION, vol.130, no.7, pp.3885-3900, 2020 (SCI-Expanded)
- 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.

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- 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.
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- XLIX. **Mutations in Chromatin Modifier and Ephrin Signaling Genes in Vein of Galen Malformation**
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NEURON, vol.101, no.3, pp.429-447, 2019 (SCI-Expanded)
- L. **Whole-exome sequencing of cervical carcinomas identifies activating ERBB2 and PIK3CA mutations as targets for combination therapy**
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PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA, vol.116, no.45, pp.22730-22736, 2019 (SCI-Expanded)
- 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**
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ANNALS OF NEUROLOGY, vol.84, no.5, pp.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, vol.103, no.5, pp.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, pp.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, vol.50, no.8, pp.1093-1107, 2018 (SCI-Expanded)
- LVI. ***De Novo* Mutation in Genes Regulating Neural Stem Cell Fate in Human Congenital Hydrocephalus**
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- LVII. **Integrated genomic analyses of de novo pathways underlying atypical meningiomas.**
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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, vol.6, no.2, pp.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, vol.4, no.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, vol.49, no.11, pp.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, vol.20, no.10, pp.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**
Yilmaz B., Toktas Z. O., Akakin A., Isik S., Bilguvar K., Kilic T., Gunel M.
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- 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.
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