Prof. Sibel Aylin UĞUR İŞERİ

Personal Information

Office Phone: +90 212 414 2000 Extension: 33314
Email: sibel.ugur@istanbul.edu.tr
Web: http://aves.istanbul.edu.tr/224/
Address: İstanbul Üniversitesi Aziz Sancar Deneysel Tıp Araştırma Enstitüsü Genetik Anabilim Dalı Vakıf Gureba Cad. Şehremi 34093, İstanbul

International Researcher IDs
ORCID: 0000-0002-5790-6853
Publons / Web Of Science ResearcherID: AAD-6099-2020
Yoksis Researcher ID: 129285

Education Information

Doctorate, Bogazici University, Institute Of Scıence, Moleküler Biyoloji Ve Genetik, Turkey 2002 - 2008
Postgraduate, Bogazici University, Institute Of Scıence, Moleküler Biyoloji Ve Genetik, Turkey 2000 - 2002
Undergraduate, Bogazici University, Fen Edebiyat Fakültesi, Moleküler Biyoloji Ve Genetik, Turkey 1996 - 2000

Foreign Languages

English, C1 Advanced

Certificates, Courses and Trainings

Education Management and Planning, Avrupa Birliği Projeleri Proje Yönetimi Sertifikası (Zertifizierter EU Project Manager 2013) European Certification and Qualification Association (Avusturya), AB Ofisi, 2013

Dissertations

Doctorate, GENOM TARAMASINDAN HASTALIK GENİ TANIMLANMASINA, Bogazici University, Fen Bilimleri Enstitüsü, Moleküler Biyoloji Ve Genetik, 2008
Postgraduate, BEŞ KALITSAL HASTALIKTA LOKUS VE GEN ANALİZI, Bogazici University, Fen Bilimleri Enstitüsü, Moleküler Biyoloji Ve Genetik, 2002

Research Areas

Life Sciences, Bioinformatics, Molecular Biology and Genetics, Genetic Disorders, Genomics, Neurobiology, Natural Sciences

Academic Titles / Tasks

Professor, Istanbul University, Aziz Sancar Institute of Experimental Medicine, Department of Genetics, 2019 - Continues
Associate Professor, Istanbul University, Aziz Sancar Institute of Experimental Medicine, Department of Genetics, 2013 - 2019
Research Assistant PhD, University of Oxford, Department Of Physiology, Anatomy And Genetics, Department Of Physiology, Anatomy And Genetics, 2008 - 2009
Research Assistant, Bogazici University, Institute Of Science, Moleküler Biyoloji Ve Genetik, 2002 - 2008

Courses

Genetics, Undergraduate, 2019 - 2020
Molecular Evolution and Biodiversity, Undergraduate, 2019 - 2020
Human Genetics, Doctorate, 2019 - 2020
İleri Moleküler Genetik, Doctorate, 2018 - 2019
Moleküler Genetik, Postgraduate, 2018 - 2019
Bioinformatics for Health Research (Biyo-Sağlık Bilişimi İngilizce Yüksek Lisans Programı) , Postgraduate, 2018 - 2019
Bioinformatics (İngilizce), Postgraduate, 2009 - 2010

Advising Theses

UĞUR İŞERİ S. A., Progresif Miyoklonik Epilepsilerde Tüm Ekzom Dizileme Yöntemi ile İlişkili Genlerinin Araştırılması, Postgraduate, G.Haryanyan(Student), 2019
UĞUR İŞERİ S. A., Zihinsel yetmezlik ile ilişkili yeni gen varyantlarının araştırılması, Doctorate, F.Yeşim(Student), 2019
UĞUR İŞERİ S. A., Beyinde Demir Birikimi ile Nörodejenerasyon Hastalığından Sorumlu Yeni Gen Varyantlarının Arsatırılması, Doctorate, N.Hande(Student), 2019
UĞUR İŞERİ S. A., GLUT1 Eksikliği Sendromunda Genetik Analizler, Postgraduate, C.Örnek(Student), 2018
UĞUR İŞERİ S. A., Febril Nöbet Sonrası Epileptogenez Sürecinin Araştırılması, Doctorate, Ö.Özdemir(Student), 2017
UĞUR İŞERİ S. A., Ailevi Epilepsilerde Tüm Genom Analizleri, Doctorate, E.Yücesan(Student), 2016
UĞUR İŞERİ S. A., İnsan genomundaki kopya sayısı değişikliklerinin SNP array yöntemi kullanılarak incelenmesi ve tespit edilen değişikliklerin validasyonu, Postgraduate, F.Yeşim(Student), 2014

Published journal articles indexed by SCI, SSCI, and AHCI

I. **Two rare autosomal recessive neurological disorders identified by combined genetic approaches in a single consanguineous family with multiple offspring.**
Susgun S., YÜCESAN E., GÖNCÜ B. S., Hasanoglu Sayin S., KINA Ü. Y., ÖZGÜL C., Duzenli O. F., Kocaturk O., Calik M., Ozbek U., et al.
Neurological sciences : official journal of the Italian Neurological Society and of the Italian Society of Clinical Neurophysiology, 2023 (SCI-Expanded)

II. **Genome-wide identification and phenotypic characterization of seizure-associated copy number variations in 741,075 individuals.**
Montanucci L., Collins R. L., Niestroj L., Parthasarathy S., Xian J., Ganesan S., Macnee M., Brünger T., Thomas R. H., Talkowski M., et al.
Nature communications, vol.14, no.1, pp.4392, 2023 (SCI-Expanded)

III. **Reanalysis of exome sequencing data reveals a treatable neurometabolic origin in two previously undiagnosed siblings with neurodevelopmental disorder**
Susgun S., Kesim Y., Khalilov D., Sirin N. G., Gezegen H., Salman B., Yucesan E., Gokcay G. F., Korbeyli H. K., Balci M. C., et al.
NEUROLOGICAL SCIENCES, vol.44, no.7, pp.2527-2540, 2023 (SCI-Expanded)

IV. **An Extraordinary EEG Phenomenon Misdiagnosed as Nonconvulsive Status Epilepticus: Frequent Subclinical Periodic Discharges Terminated by Sudden Auditory Stimuli.**
Oguz-Akarsu E., Salman B., Ugur-Iseri S. A., Baykan B.
Clinical EEG and neuroscience, vol.54, no.2, pp.160-163, 2023 (SCI-Expanded)

V. **Obstacles and expectations of rare disease patients and their families in Turkiye: ISTisNA project survey results**
HATIRNAZ NG Ö., Sahin I., ERBİLGİN Y., ÖZDEMİR Ö., YÜCESAN E., Erturk N., Yemenici M., AKGÜN DOĞAN Ö., UĞUR İŞERİ S. A., SATMAN İ., et al.
FRONTIERS IN PUBLIC HEALTH, vol.10, 2023 (SCI-Expanded)

VI. **Clinical and genetic analyses in syndromic intellectual disability with primary microcephaly reveal biallelic and de novo variants in patients with parental consanguinity.**
Mercan S., Akcakaya N. H., Salman B., Yapici Z., Ozbek U., Ugur Iseri S. A.
Genes & genomics, vol.45, no.1, pp.13-21, 2023 (SCI-Expanded)

VII. **Targeted resequencing reveals high-level mosaicism for a novel frameshift variant in WDR45 associated with beta-propeller protein-associated neurodegeneration**
Susgun S., DEMİREL M., YALÇIN ÇAKMAKLI G., Salman B., Oguz K. K., ELİBOL B., UĞUR İŞERİ S. A., Yapici Z.
International Journal of Neuroscience, 2023 (SCI-Expanded)

VIII. **Epilepsy or neurodevelopmental disorders are associated with homozygous and pathogenic ELP2 variation in three siblings.**
Khalilov D., Haryanyan G., Salman B., YÜCESAN E., Ugur Iseri S. A., Bebek N.
Neurocase, vol.28, no.6, pp.488-492, 2022 (SCI-Expanded)

IX. **Quantitative Measurement of Iron in NBIA patients with Quantitative Susceptibility Mapping and Clinical Evaluation**
Uygun O., Iseri S. U., Ozcan A., DİNÇER A., Yapici Z.
EUROPEAN JOURNAL OF NEUROLOGY, vol.29, pp.626, 2022 (SCI-Expanded)

X. **Hypomyelinating spastic dyskinesia and ichthyosis caused by a homozygous splice site mutation leading to exon skipping in ELOVL1.**
Takahashi T., Mercan S., Sassa T., BAYRAM AKÇAPINAR G., Yararbas K., SÜSGÜN S., Iseri S. A., Kihara A., Akcakaya N. H.
Brain & development, vol.44, no.6, pp.391-400, 2022 (SCI-Expanded)

XI. **Two cases with mitochondrial membrane protein-associated neurodegeneration: genetic features and long-term clinical follow-up.**
Mercan S., Ugur Iseri S. A., Yigiter R., Akcakaya N. H., Saka E., Yapici Z.
Neurocase, vol.28, no.1, pp.37-41, 2022 (SCI-Expanded)

XII. **The rare rs769301934 variant in NHLRC1 is a common cause of Lafora disease in Turkey.**
Haryanyan G., Ozdemir O., Tutkavul K., Dervent A., Ayta S., ÖZKARA Ç., Salman B., YÜCESAN E., Kesim Y., Susgun S., et al.
Journal of human genetics, vol.66, no.12, pp.1145-1151, 2021 (SCI-Expanded)

XIII. **Distinct gene-set burden patterns underlie common generalized and focal epilepsies**
Koko M., Krause R., Sander T., Bobbili D. R., Nothnagel M., May P., Lerche H.
EBIOMEDICINE, vol.72, 2021 (SCI-Expanded)

XIV. **Shared genetic basis between genetic generalized epilepsy and background electroencephalographic oscillations**
Stevelink R., Luykx J. J., Lin B. D., Leu C., Lal D., Smith A. W., Schijven D., Carpay J. A., Rademaker K., Baldez R. A. R., et al.
EPILEPSIA, vol.62, no.7, pp.1518-1527, 2021 (SCI-Expanded)

XV. **Sub-genic intolerance, ClinVar, and the epilepsies: A whole-exome sequencing study of 29,165 individuals**
Motelow J. E., Povysil G., Dhindsa R. S., Stanley K. E., Allen A. S., Feng Y. A., Howrigan D. P., Abbott L. E., Tashman K., Cerrato F., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, no.6, pp.965-982, 2021 (SCI-Expanded)

XVI. **Effect of the brain-derived neurotrophic factor gene Val66Met polymorphism on sensory-motor integration during a complex motor learning exercise.**
Deveci S., Matur Z., Kesim Y., Senturk Ş., Sargın-Kurt G., Ugur U., Oge Ö.
Brain research, vol.1732, pp.146652, 2020 (SCI-Expanded)

XVII. **Polygenic burden in focal and generalized epilepsies.**
Leu C., Stevelink R., Smith A. W., Goleva S. B., Kanai M., Ferguson L., Campbell C., Kamatani Y., Okada Y., Sisodiya S. M., et al.
Brain : a journal of neurology, vol.142, no.11, pp.3473-3481, 2019 (SCI-Expanded)

XVIII. **Ultra-Rare Genetic Variation in the Epilepsies: A Whole-Exome Sequencing Study of 17,606 Individuals**
Feng Y. A., Howrigan D. P., Abbott L. E., Tashman K., Cerrato F., Singh T., Heyne H., Byrnes A., Churchhouse C., Watts N., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, no.2, pp.267-282, 2019 (SCI-Expanded)

XIX. **Biallelic loss of EEF1D function links heat shock response pathway to autosomal recessive intellectual disability**
Iseri S. A., YÜCESAN E., Tuncer F. N., Calik M., Kesim Y., Uzun G. A., Ozbek U.
JOURNAL OF HUMAN GENETICS, vol.64, no.5, pp.421-426, 2019 (SCI-Expanded)

XX. **A Novel and Mosaic WDR45 Nonsense Variant Causes Beta-Propeller Protein-Associated Neurodegeneration Identified Through Whole Exome Sequencing and X chromosome Heterozygosity Analysis.**
Akcakaya N. H., Salman B., Gormez Z., Arguden Y. T., Cirakoglu A., Cakmur R., DÖNMEZ ÇOLAKOĞLU B., Hacihanefioglu S., ÖZBEK U., Yapici Z., et al.
Neuromolecular medicine, vol.21, no.1, pp.54-59, 2019 (SCI-Expanded)

XXI. **Identification of epilepsy related pathways using genome-wide DNA methylation measures: A trio-based approach**
OZDEMIR O., EGEMEN E., Ugur I., SEZERMAN O., Bebek N., Baykan B., Ozbek U.
PLOS ONE, vol.14, no.2, 2019 (SCI-Expanded)

XXII. **Clinical and genetic spectrum of an orphan disease MPAN: a series with new variants and a novel phenotype.**
Akcakaya N. H., Haryanyan G., Mercan S., Sozer N., Ali A., Tombul T., ÖZBEK U., Iseri S. A., Yapici Z.
Neurologia i neurochirurgia polska, vol.53, no.6, pp.476-483, 2019 (SCI-Expanded)

XXIII. **A novel homozygous GALC variant has been associated with Krabbe disease in a consanguineous family**
Tuncer F. N., Iseri S. A., Yapici Z., Demir M., Karaca M., Calik M.
NEUROLOGICAL SCIENCES, vol.39, no.12, pp.2123-2128, 2018 (SCI-Expanded)

XXIV. **Investigation of SLC2A1 gene variants in genetic generalized epilepsy patients with eyelid myoclonia**
ALTıOKKA-UZUN G., OZDEMIR O., Uğur-İşeri S. A., Bebek N., Gürses C., Özbek U., Baykan B.
EPILEPTIC DISORDERS, vol.20, no.5, pp.396-400, 2018 (SCI-Expanded)

XXV. **Clinical phenotype of hereditary spastic paraplegia due to KIF1C gene mutations across life span.**
Yucel-Yilmaz D., Yucesan E., YALNIZOĞLU D., Oguz K. K., SAGIROGLU M. S., Ozbek U., Serdaroglu E., Bilgic B., Erdem S., Iseri S. A., et al.
Brain & development, vol.40, no.6, pp.458-464, 2018 (SCI-Expanded)

XXVI. **Third-line treatment with second-generation tyrosine kinase inhibitors (dasatinib or nilotinib) in patients with chronic myeloid leukemia after two prior TKIs: real-life data on a single center experience along with the review of the literature.**
Ongoren S., Eskazan A. E., Suzan V., Savci S., Ozunal I. E., Berk S., Yalniz F. F., Elverdi T., Salihoglu A., Erbilgin Y., et al.
Hematology (Amsterdam, Netherlands), vol.23, no.4, pp.212-220, 2018 (SCI-Expanded)

XXVII. **SYNE1 related cerebellar ataxia presents with variable phenotypes in a consanguineous family from Turkey**
Yucesan E., UĞUR İŞERİ S. A., Bilgic B., Gormez Z., Bakir Gungor B., Sarac A., ÖZDEMİR Ö., Sagiroglu M., Gurvit H., Hanagasi H. A., et al.

NEUROLOGICAL SCIENCES, vol.38, no.12, pp.2203-2207, 2017 (SCI-Expanded)

XXVIII. **Clinical and genetic features of PKAN patients in a tertiary centre in Turkey**
AKÇAKAYA N., Iseri S. A., BILIR B., BATTALOGLU E., TEKTURK P., GULTEKIN M. H., AKAR G., YIGITER R., Hanagasi H. A., ALP R., et al.
CLINICAL NEUROLOGY AND NEUROSURGERY, pp.34-42, 2017 (SCI-Expanded)

XXIX. **Identification and functional characterisation of genetic variants in OLFM2 in children with developmental eye disorders**
Holt R., Iseri S. A., Wyatt A. W., Bax D. A., Diaz D. G., Santos C., Broadgate S., Dunn R., Bruty J., Wallis Y., et al.
HUMAN GENETICS, vol.136, no.1, pp.119-127, 2017 (SCI-Expanded)

XXX. **Differential Diagnosis of Bithalamic and Pallidal Hypointensity - a Case of HEXB Mutation**
Akcakaya N. H., Ozdemir O., Gokcay F. G., Iseri S. A., Yapici Z.
CESKA A SLOVENSKA NEUROLOGIE A NEUROCHIRURGIE, vol.80, no.3, pp.343-345, 2017 (SCI-Expanded)

XXXI. **A novel gene mutation in PANK2 in a patient with severe jaw-opening dystonia**
Yapici Z., Akcakaya N. H., Tekturk P., Iseri S. A., Ozbek U.
BRAIN & DEVELOPMENT, no.8, pp.755-758, 2016 (SCI-Expanded)

XXXII. **Screening LGI1 in a cohort of 26 lateral temporal lobe epilepsy patients with auditory aura from Turkey detects a novel de novo mutation**
Kesim Y. F., Uzun G. A., Yucesan E., Tuncer F. N., Ozdemir O., Bebek N., Ozbek U., Iseri S. A., Baykan B.
EPILEPSY RESEARCH, vol.120, pp.73-78, 2016 (SCI-Expanded)

XXXIII. **A clinical variant in SCN1A inherited from a mosaic father cosegregates with a novel variant to cause Dravet syndrome in a consanguineous family**
Tuncer F. N., Görmez Z., Calik M., Uzun G. A., SAGIROGLU M. S., YUCETURK B., YUKSEL B., Baykan B., Bebek N., İŞCAN A., et al.
EPILEPSY RESEARCH, vol.113, pp.5-10, 2015 (SCI-Expanded)

XXXIV. **Identifying SNP targeted pathways in partial epilepsies with genome-wide association study data**
Bakir-Gungor B., Baykan B., Iseri S. U., Tuncer F. N., Sezerman O. U.
EPILEPSY RESEARCH, vol.105, pp.92-102, 2013 (SCI-Expanded)

XXXV. **A frameshift mutation of ERLIN2 in recessive intellectual disability, motor dysfunction and multiple joint contractures**
Yildirim Y., Orhan E., Iseri S. A. U., Serdaroglu-Oflazer P., KARA B., Solakoglu S., Tolun A.
HUMAN MOLECULAR GENETICS, vol.20, no.10, pp.1886-1892, 2011 (SCI-Expanded)

XXXVI. **Early-Onset Progressive Myoclonic Epilepsy With Dystonia Mapping to 16pter-p13.3**
Duru N., UĞUR İŞERİ S. A., Selcuk N., TOLUN A.
JOURNAL OF NEUROGENETICS, vol.24, no.4, pp.207-215, 2010 (SCI-Expanded)

XXXVII. **A novel recessive GUCY2D mutation causing cone-rod dystrophy and not Leber's congenital amaurosis.**
Ugur I., DURLU Y., TOLUN A.
European journal of human genetics : EJHG, vol.18, pp.1121-6, 2010 (SCI-Expanded)

XXXVIII. **Use of genome-wide SNP homozygosity mapping in small pedigrees to identify new mutations in VSX2 causing recessive microphthalmia and a semidominant inner retinal dystrophy**
Iseri S. A., Wyatt A. W., NUERNBERG G., KLUCK C., NUERNBERG P., Holder G. E., Blair E., Salt A., Ragge N. K.
HUMAN GENETICS, vol.128, no.1, pp.51-60, 2010 (SCI-Expanded)

XXXIX. **Sonic Hedgehog Mutations Are an Uncommon Cause of Developmental Eye Anomalies**
Bakrania P., Iseri S. A., Wyatt A. W., BUNYAN D. J., Lam W. W. K., Salt A., Ramsay J., ROBINSON D. O., Ragge N. K.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, no.5, pp.1310-1313, 2010 (SCI-Expanded)

XL. **Seeing Clearly: The Dominant and Recessive Nature of FOXE3 in Eye Developmental Anomalies**
Iseri S. A., Osborne R. J., Farrall M., Wyatt A. W., Mirza G., NUERNBERG G., KLUCK C., HERBERT H., Martin A., HUSSAIN M. S., et al.
HUMAN MUTATION, vol.30, no.10, pp.1378-1386, 2009 (SCI-Expanded)

XLI. **Very-late-onset pyridoxine-dependent seizures not linking to the known 5q31 locus.**
KABAKUS N., AYDIN M., Ugur S. A., DURUKAN M., TOLUN A.

Pediatrics international : official journal of the Japan Pediatric Society, vol.50, pp.703-5, 2008 (SCI-Expanded)

XLII. **Homozygous WNT10b mutation and complex inheritance in Split-Hand/Foot Malformation**
Ugur S. A., Tolun A.
HUMAN MOLECULAR GENETICS, vol.17, no.17, pp.2644-2653, 2008 (SCI-Expanded)

XLIII. **A deletion in DRCTNNB1A associated with hypomyelination and juvenile onset cataract**
Ugur S. A., Tolun A.
EUROPEAN JOURNAL OF HUMAN GENETICS, vol.16, no.2, pp.261-264, 2008 (SCI-Expanded)

XLIV. **Mutations in SLC34A2 cause pulmonary alveolar microlithiasis and are possibly associated with testicular microlithiasis**
CORUT A., SENYIGIT A., Ugur S. A., ALTIN S., OZCELIK U., CALISIR H., YILDIRIM Z., GOCMEN A., TOLUN A.
AMERICAN JOURNAL OF HUMAN GENETICS, vol.79, no.4, pp.650-656, 2006 (SCI-Expanded)

XLV. **Severe form of Cockayne syndrome with varying clinical presentation and no photosensitivity in a family.**
SONMEZ F., CELEP F., Ugur S. A., TOLUN A.
Journal of child neurology, vol.21, pp.333-7, 2006 (SCI-Expanded)

XLVI. **Identification of a locus for an autosomal recessive hyaline body myopathy at chromosome 3p22.2-p21.32.**
ONENGÜT S., Uğur S. A., KARASOY H., YÜCEYAR N. S., TOLUN A.
Neuromuscular disorders : NMD, vol.14, pp.4-9, 2004 (SCI-Expanded)

XLVII. **Is the novel SCKL3 at 14q23 the predominant Seckel locus?**
KILINC M., NINIS V., Ugur S. A., Tuysuz B., Seven M., BALCI S., GOODSHIP J., TOLUN A.
EUROPEAN JOURNAL OF HUMAN GENETICS, vol.11, no.11, pp.851-857, 2003 (SCI-Expanded)

Articles Published in Other Journals

I. **COMBINED ANALYSIS OF LINKAGE AND WHOLE EXOME SEQUENCING REVEALS CIC AS A CANDIDATE GENE FOR ISOLATED DYSTONIA**
Salman B., YÜCESAN E., SAMANCI B., BİLGİÇ B., HANAĞASI H. A., Gurvit I., ÖZBEK U., UĞUR İŞERİ S. A.
JOURNAL OF ISTANBUL FACULTY OF MEDICINE-ISTANBUL TIP FAKULTESI DERGISI, vol.84, no.4, pp.457-463, 2021 (ESCI)

II. **SCREENING SLC2A1 GENE FOR SEQUENCE AND COPY NUMBER VARIATIONS ASSOCIATED WITH GLUT-1 DEFICIENCY SYNDROME**
Ornek Erguzeloglu C., KARA B., Karacan I., Ozdemir O., Kesim Y., Bebek N., Ozbek U., Ugur Iseri S. A.
JOURNAL OF ISTANBUL FACULTY OF MEDICINE-ISTANBUL TIP FAKULTESI DERGISI, vol.83, no.3, pp.177-183, 2020 (ESCI)

III. **SCREENING SLC2A1 GENE FOR SEQUENCE AND COPY NUMBER VARIATIONS ASSOCİATED WITH GLUT-1 DEFICIENCY SYNDROME**
Örnek Ergüzeloğlu C., Kara B., Karacan İ., Özdemir Ö., Kesim Y., BEBEK N., Özbek U., UĞUR İŞERİ S. A.
JOURNAL OF ISTANBUL FACULTY OF MEDICINE-ISTANBUL TIP FAKULTESI DERGISI, vol.21, pp.1-7, 2019 (Peer-Reviewed Journal)

IV. **Kopya Sayısı Değişikliklerinin SNP Array ile Tespiti- Detection of Copy Number Variations using SNP arrays**
UĞUR İŞERİ S. A.
Türkiye Klinikleri J Pediatr Sci., vol.12, no.4, pp.54-59, 2016 (Peer-Reviewed Journal)

V. **Tüm Genom SNP Genotipleme ile Trizomi ve Ebeveyn Etkisinin Tespiti**
Kesim F. Y., TUNCER KILINÇ F. N., YUCESAN E., OZDEMIR O., CALIK M., ÖZBEK U., UĞUR İŞERİ S. A.
Deneysel Tıp Degisi, vol.3, no.7, pp.22-27, 2015 (Peer-Reviewed Journal)

VI. **Tüm Genom SNP Genotipleme ile Trizomi ve Ebeveyn Etkisinin Tespiti**
Kesim F. Y., TUNCER KILINÇ F. N., YUCESAN E., OZDEMIR O., CALIK M., ÖZBEK U., UĞUR İŞERİ S. A.
Deneysel Tıp Dergisi, vol.3, no.7, pp.22-27, 2014 (Peer-Reviewed Journal)

VII. **Nörogenetikte bağlantı analizinin kullanımı**
UĞUR İŞERİ S. A., ÖZBEK U.
Türkiye Klinikleri J Neurol-Special Topics, vol.4, no.2, pp.11-15, 2011 (Peer-Reviewed Journal)

Books & Book Chapters

I. **Nadir Nörolojik Hastalıklarda Gen Avcılığı**
Uğur İşeri S. A.
in: Türkiye Klinikleri Tıbbi Genetik - Özel Konular, Prof. Dr. Uğur ÖZBEK, Editor, Türkiye Klinikleri Yayınevi, İstanbul, pp.56-63, 2021

II. **Biyobankalar**
Erbilgin Y., Uğur İşeri S. A., Özbek U.
in: Tıp Bilişimi, Nilgün Bozbuğa,Sevinç Gülseçen, Editor, Istanbul University, İstanbul, pp.159-169, 2021

III. **Biyobankalamada Protokol ve Kalite Kılavuzu**
ERBİLGİN Y., UĞUR İŞERİ S. A., Khodzhaev K., Haryanyan G.
İstanbul Medikal Sağlık ve Yayıncılık Hiz. Tic. Ltd. Şti., İstanbul, 2017

Refereed Congress / Symposium Publications in Proceedings

I. **Nadir Nörogenetik Bir Hastalik Modeli Olarak Epileptik Ensefelopatilerde Trio Ekzom Dizileme Analizleri**
Karaaslan Z., Salman B., Ovamigyan S. A., Kılıç M., Çırak S., Uğur İşeri S. A., Yılmaz V., Tüzün E., Küçükali C. İ., Yapıcı Z., et al.
59. Ulusal Nöroloji Kongresi, Antalya, Turkey, 13 - 18 December 2023, pp.25

II. **Multiple Rating Scales in 17 patients with Mitochondrial-membrane Protein Associated Neurodegeneration**
SAYMAN C., ÇAPAN N., TOPALOĞLU P., UĞUR İŞERİ S. A., ÖZDEMİR S., BASLO M. B., KOCASOY ORHAN E., AYDIN A. R., YAPICI OBUZ Z.
17th Congress of the European Forum for Research in Rehabilitation, 02 November 2023

III. **KRONİK MİYELOİD LÖSEMİ HASTALARINDA FARKLI BCR::ABL1 TRANSKRİPTLERİNİN KLİNİK YANSIMALARI**
Ordu M., Erbilgin Y., Yılmaz U., Eşkazan A. E., Altındirek D., Uğur İşeri S. A., Hatırnaz Ng Ö., Özbek U., Sayitoğlu M.
48. Ulusal Hematoloji Kongresi, Antalya, Turkey, 1 - 05 November 2022, pp.126-127

IV. **Kronik Miyeloid Lösemi Hastalarında Farklı BCR::ABL1 Transkriptlerinin Klinik Yansımaları**
Ordu M., ERBİLGİN Y., YILMAZ U., EŞKAZAN A. E., UĞUR İŞERİ S. A., HATIRNAZ NG Ö., ÖZBEK U., SOYSAL T., SAYİTOĞLU M.
48. Ulusal Hematoloji Kongresi, Antalya, Turkey, 1 - 05 November 2022

V. **Outcomes of a Biobanking Initiative from Turkey: 'Investing in the Future: BIOBANK**
ERBİLGİN Y., TUNCER F. N., UĞUR İŞERİ S. A., Khodzhaev K., ÖZBEK U.
Global Biobank Week, 13 - 15 September 2017

VI. **Towards an Epilepsy Biobank**
Haryanyan G., Özdemir Ö., Yücesan E., TUNCER F. N., Kesim F. Y., ERBİLGİN Y., Khodzhaev K., UĞUR İŞERİ S. A., ÖZKARA Ç., BAYKAL B., et al.
Global Biobank Week, 13 - 15 September 2017

VII. **Investing in the Future BIOBANK**
ERBİLGİN Y., TUNCER F. N., Haryanyan G., Khodzhaev K., UĞUR İŞERİ S. A., ÖZBEK U.
European Biobank Week 2016, 13 - 16 September 2016

VIII. **Exome Sequencing Combined with Linkage Analysis Identifies a Novel Gene Associated with a Syndromic Form of Intellectual Disability**

Kesim Y., Çalık M., Tuncer F., İşcan A., Uzun G., Özbek U., UĞUR İŞERİ S. A.
European Society of Human Genetics 2016, Barselona, Spain, 21 - 26 May 2016, pp.175

IX. **Glut1 eksikliği sendromu klinik fenotipleri**
KARA B., ÖZDEMİR Ö., MARAŞ GENÇ H., UYUR YALÇIN E., SAKARYA GÜNEŞ A., UĞUR İŞERİ S.
18. ULUSAL ÇOCUK NÖROLOJİ KONGRESİ, Antalya, Turkey, 20 April 2016

X. **Correlation between phenotypic and genotypic tetracycline resistance of Escherichia coli isolates from food of animal origin**
Muratoglu K., Özdemir Ö., Eker F. Y., Bayrakal M., Levent G., Ugur S. A., Ozbek U., Ciftcioglu G.
European Biotechnology Congress, Lecce, Italy, 15 - 18 May 2014, vol.185

Other Publications

I. **Febril nöbet sonrası epileptogenez sürecinin araştırılması**
Özdemir Ö., Uğur Işeri S. A.
Other, pp.1-100, 2017

Supported Projects

UĞUR İŞERİ S. A., Other International Funding Programs, Epi25 collaborative: Large-Scale Whole Genome Sequencing in Epilepsy, 2017 - Continues
UĞUR İŞERİ S. A., Project Supported by Higher Education Institutions, Zihinsel Yetmezlik ile İlişkili Yeni Gen Varyantlarının Araştırılması, 2017 - 2019
TÜRKER ŞENER L., UĞUR İŞERİ S. A., TUNCER KILINÇ F. N., ERBİLGİN Y., Project Supported by Other Official Institutions, Geleceğe Yatırım: BİYOBANKA, 2015 - 2017
YAPICI OBUZ Z., HANAĞASI H. A., UĞUR İŞERİ S. A., Project Supported by Higher Education Institutions, Beyinde demir birikimi ile nörodejenerasyonu olan hastaların genetik analizi, 2015 - 2017
TUNCER KILINÇ F. N., UĞUR İŞERİ S. A., ÖZBEK U., KARA B., MARAŞ GENÇ H., UYUR YALÇIN E., Project Supported by Higher Education Institutions, A novel mutation in PCDH19 enabled genetic diagnosis as Epilepsy and Mental Retardation Limited to Females, 2016 - 2016
ÖZBEK U., UĞUR İŞERİ S. A., HANAĞASI H. A., CEYLAN N. H., YAPICI Z., ERAKSOY M., TEKTÜRK P., BATTALOĞLU E., Project Supported by Higher Education Institutions, Clinical and Genetic Features of PKAN Patients in a Tertiary Center in Turkey, 2016 - 2016
UĞUR İŞERİ S. A., İŞCAN A., ÖZBEK U., ÇALIK M., KESİM F. Y., TUNCER F. N., Project Supported by Higher Education Institutions, Exome Sequencing Combined with Linkage Analysis Identifies a Novel Gene Associated with a Syndromic Form of Intellectual Disability, 2016 - 2016
UĞUR İŞERİ S. A., Project Supported by Higher Education Institutions, İdyopatik Jeneralize Epilepsi ile İlişkili rs113904207 İsimli Nadir Varyantın Sıklığının Tespiti, 2014 - 2015
UĞUR İŞERİ S. A., TUBITAK Project, Nadir Epilepsi Sendromları Genetiği, 2011 - 2014
TUBITAK Project, İdyopatik Jeneralize Epilepsilerde Tüm Genom Metilom Analizi, 2010 - 2013
UĞUR İŞERİ S. A., EU Framework Program Project, EPICURE: Functional Genomics and Neurobiology of Epilepsy: a basis for new therapeutic strategies, 2007 - 2011
UĞUR İŞERİ S. A., Other International Funding Programs, Novel approaches for the genetic diagnosis of eye developmental disorders-NewLife Foundation for Disabled Children. İngiltere, 2008 - 2009
UĞUR İŞERİ S. A., Other International Funding Programs, Monogenik hastalıklar için 400 bireyde genotipleme - NHLBI Mammalian Genotyping Service, Marshfield Clinic Research Foundation, 2002 - 2008
UĞUR İŞERİ S. A., CB Strateji ve Bütçe Başkanlığı (Kalkınma Bakanlığı) Projesi, Nadir hastalık genleri belirlenmesi, 2004 - 2004

Memberships / Tasks in Scientific Organizations

İstanbul Teknik Üniversitesi Sağlık ve Mühendislik Bilimleri İnsan deneyleri (SM-INAREK) Etik Kurulu, Member, 2020 - Continues, Turkey
İstanbul Üniversitesi Epilepsi Uygulama ve Araştırma Merkezi (EPİMER) Yönetim Kurulu Üyesi, Board Member, 2018 - Continues
European Society of Human Genetics, Member, 2016 - Continues
American Society of Human Genetics, Member, 2016 - Continues
Türk Nöroloji Derneği, Member, 2014 - Continues

Metrics

Publication: 68
Citation (WoS): 765
Citation (Scopus): 845
H-Index (WoS): 14
H-Index (Scopus): 14

Congress and Symposium Activities

ZIHINSEL OGRENME YETERSIZLİGININ ESLIK ETTIGI MIKROSEFALI OLGULARINDA TU¨M EKZOM DIZILEME UYGULAMALARI. 13. Ulusal Tıbbi Genetik Kongresi., Attendee, Antalya, Turkey, 2018
LATERAL TEMPORAL LOB EPİLEPSİLİ HASTALARDA GENETIK ANALIZLER. 13. Ulusal Tıbbi genetik Kongresi., Attendee, Turkey, 2018
EPM4 ILE ILISKILI YENI SCARB2 VARYASYONUNUN TUM EKZOM DIZILEME YONTEMI ILE TESPITI. 11. Ulusal Epilepsi Kongresi., Attendee, Turkey, 2018
114 kisilik epilepsi kohortunda ekzom dizileme verisi ile SCN1A gen varyantlarının meta analizi. 11. Ulusal Epilepsi Kongresi., Attendee, Muğla, Turkey, 2018
GLUT1 EKSIKLIGI SENDROMUNDA SLC2A1 GENI KOPYA SAYISI DEGISIKLIKLERININ INCELENMESI. 11. Ulusal Epilepsi Kongresi., Attendee, Turkey, 2018
Exome sequencing identifies a novel FBXO38 variant inherited from a mosaic mother to cause Distal Hereditary Motor Neuronopathy Type IID with distinct features. 67th Annual Meeting of the American Society of Human Genetics, Attendee, Florida, United States Of America, 2017
Research Biobank for Leukemia.Global Biobank Week 2017, Attendee, Stockholm, Sweden, 2017
Towards an Epilepsy Biobank. Global Biobank Week 2017, Attendee, Stockholm, Sweden, 2017
Outcomes of a Biobanking Initiative from Turkey: 'Investing in the Future: BIOBANK'., Attendee, Stockholm, Sweden, 2017
EPILEPSININ ESLIK ETTIGI ZIHINSEL YETMEZLIK VAKASINDA GENOMIK YAKLASIMLAR. 53. Ulusal Nöroloji Kongresi., Attendee, Antalya, Turkey, 2016
GLUT1 eksikligi sendromunda genetik analizler. 53. Ulusal Nöroloji Kongresi., Attendee, Turkey, 2016
SENDROMIK ZIHINSEL YETMEZLIKTE TESPIT EDILEN YENI BIR GEN VE IN VITRO KARAKTERIZASYONU. 53. Ulusal Nöroloji Kongresi., Attendee, Turkey, 2016
Febril nöbet sonrası epoleptogenez sürecinin araştırılması. 53. Ulusal Nöroloji Kongresi., Attendee, Turkey, 2016
Investing in the Future BIOBANK. Europe Biobank Week 2016, Attendee, Vienna, Austria, 2016
Clinical and Genetic Features of PKAN Patients in a Tertiary Center in Turkey. European Human Genetics Conference 2016, Attendee, Barcelona, Spain, 2016
BENIGN FAMILYAL INFANTIL KONVULSIYONA SAHIP BIREYLERI OLAN GENIS BIR AILEDE PRRT2 GEN ANALIZININ GERCEKLESTIRILMESI. 10. Ulusal Epilepsi Kongresi., Attendee, Turkey, 2016
GLUT1 YETMEZLIK SENDROMU ILE ILISKILI SLC2A1 DE NOVO GEN VARYANTLARININ TESPITI. 10. Ulusal Epilepsi

Kongresi., Attendee, Turkey, 2016
FEBRIL NOBET SONRASI EPILEPTOGENEZ SURECININ GENOM BOYU ANALIZLERLE SICAN MODELINDE ARASTIRILMASI (GENOME-WIDE ANALYSIS OF EPILEPTOGENESIS ON FEBRILE SEIZURE RAT MODEL). 10. Ulusal Epilepsi Kongresi., Attendee, Turkey, 2016
LATERAL TEMPORAL LOB EPILEPSI HASTALARINDA GERCEKLESTIRILEN LGI1 TARAMASINDA YENI BIR DE NOVO MUTASYON TESPITI. 10. Ulusal Epilepsi Kongresi., Attendee, İzmir, Turkey, 2016
A novel mutation in PCDH19 enabled genetic diagnosis as Epilepsy and Mental Retardation Limited to Females. European Human Genetics Conference 2016, Attendee, Barcelona, Spain, 2016
Exome Sequencing Combined with Linkage Analysis Identifies a Novel Gene Associated with a Syndromic Form of Intellectual Disability. European Human Genetics Conference 2016, Attendee, Barcelona, Spain, 2016
Genetic analysis of delayed motor mental development and Unverricht-Lundborg disease in a large highly consanguineous family from Turkey. European Human Genetics Conference 2016, Attendee, Barcelona, Spain, 2016
Mikrosefali-lenfödem-koryoretinaldisplazi-mikrooftalmi sendromlu bir olguda KIF11 gen mutasyonu ve 22q11.2. 3. Nörometabolik Dosmorfoloji Sempozyumu., Attendee, İstanbul, Turkey, 2016
Krabbe Hastalığı: Yeni bir GALC gen varyantının tespiti ile akraba evliliği yapmış ailede ayırıcı tanının sağlanması. 3. Nörometabolik Dosmorfoloji Sempozyumu., Attendee, Turkey, 2016
RNA-SPESIFIK ADENOZIN DEAMINAZ (ADAR) GENINDE YENI BIR MUTASYONUN NEDEN OLDUGU AICARDI-GOUTIERES TIP 6 SENDROMU. 51. Ulusal Nöroloji Kongresi, Attendee, Antalya, Turkey, 2015
PANTOTENAT-KINAZ İLİŞKİLİ NÖRODEJENERASYON: KLINIK-GENETIK-RADYOLOJIK CALISMA. 51. Ulusal Nöroloji Kongresi, Attendee, Turkey, 2015
MKA/MR olgularında kopya sayısı degisimlerinin SNP- array yöntemi ile incelenmesi ve validasyonu. 12. Ulusal Çocuk Genetik Sempozyumu, Attendee, Samsun, Turkey, 2015
Akraba evliligi yapmıs¸genis¸bir ailede birbirinden bagımsız olarak gozlenen Unverricht-Lundborg Hastalıgı ve Motor Mental Gelisme Geriliginin genetik olarak incelenmesi. 12. Ulusal Çocuk Genetik Sempozyumu, Attendee, Turkey, 2015
A clinical variant in SCN1A inherited from a mosaic father cosegregates with a novel variant to cause Dravet syndrome in a consanguineous family. European Human Genetics Conference 2015, Attendee, Glasgow, United Kingdom, 2015
A novel mutation in LGI1 gene found in a series of consecutive epileptic patients with auditory aura. European Human Genetics Conference 2015, Attendee, Glasgow, United Kingdom, 2015
Combined analysis of linkage and exome sequencing identifies a novel elongation factor associated with intellectual disability and delayed motor development. European Human Genetics Conference 2015, Attendee, Glasgow, United Kingdom, 2015
Combined analysis of linkage and exome sequencing identifies a novel SYNE1 mutation in a consanguineous family from Turkey with a rare form of recessive cerebellar ataxia. European Human Genetics Conference 2014, Attendee, Milan, Italy, 2014
Whole genome methylation analysis in idiopathic generalized epilepsies. European Human Genetics Conference 2014, Attendee, Milan, Italy, 2014
Identifying disease genes in a large highly inbred consanguineous kindred from Turkey with idiopathic generalized epilepsy. American Society of Human Genetics 62nd Annual Meeting, Attendee, California, United States Of America, 2012
Genome-wide SNP analysis in a large consanguineous Turkish family reveals diagnosis as Unverricht-Lundborg Disease. American Society of Human Genetics 62nd Annual Meeting, Attendee, California, United States Of America, 2012
Identification of a de novo splice-site mutation in SLC2A1 gene causing Glut1 deficiency syndrome in a Turkish patient. European Human Genetics Conference 2012, Attendee, Nuremberg, Germany, 2012
GENETIC ANALYSIS OF SLC2A1 GENE CODING GLUCOSE TRANSPORTER GLUT1 IN TURKISH IDIOPATHIC GENERALIZED EPILEPSY PATIENTS. 29th International Epilepsy Congress, Attendee, Rome, Italy, 2011
Whole genome linkage analysis in a large consanguineous Turkish family with idiopathic generalized epilepsy. European Human Genetics Conference 2011, Attendee, Amsterdam, Netherlands, 2011
Detection of copy number events in patients with eye development disorders using SNP arrays and multiple detection algorithms. American Society of Human Genetics 59th Annual Meeting, Attendee, Hawaii, United States Of America, 2009
The 94th annual Meeting of the Oxford Ophthalmological Congress: Identification of novel mutations in a lens specific transcription factor gene, Attendee, Oxford, United Kingdom, 2009
Sonic Hedgehog Mutations are an Uncommon Cause of Developmental Eye Anomalies. European Human Genetics

Conference 2009, Attendee, Vienna, Austria, 2009
Linkage to 13q in a novel autosomal dominant pseudoarthrogryposis-like syndrome. European Human Genetics Conference 2007, Attendee, Nice, France, 2007
Mutations in a solute carrier gene are responsible for pulmonary alveolar and partly for testicular microlithiasis. 31st Congress of the Federation-of-European-Biochemical-Societies (FEBS) 2006., Attendee, İstanbul, Turkey, 2006
An early-onset progressive encephalopathy with myoclonus and dystonia (PEMD) mapping to chromosome 16pter. European Human Genetics Conference 2006, Attendee, Amsterdam, Netherlands, 2006
Locus and gene analysis in a novel autosomal recessive leukodystrophy. European Human Genetics Conference 2006, Attendee, Amsterdam, Netherlands, 2006

Scholarships

Bilim Adamı Yetiştirme Grubu Bursiyeri (BAYG/BİDEB) 2001-2006, TUBITAK, 2001 - 2006

Awards

Uğur İşeri S. A., TÜBA GEBİP 2019, Türkiye Bilimler Akademisi, December 2019
UĞUR İŞERİ S. A., Tüm genom bağlantı analizi ve aday gen incelemesi ile otozomal çekinik İJE'de genetik faktörlerin araştırılması, Beyin Araştırmaları Derneği-Boehringer-Ingelheim Proje Desteği, May 2010

Non Academic Experience

University of Oxford
Boğaziçi Üniversitesi