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Personal Information

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Education Information

Expertise In Medicine, Istanbul University, Cerrahpaşa Tıp Fakültesi, Turkey 2015 - 2019
Doctorate, Istanbul University, Health Sciences Institute, Turkey 1989 - 1994
Undergraduate, Istanbul University, Cerrahpaşa Tıp Fakültesi, Cerrahpaşa Tıp Pr., Turkey 1979 - 1986

Dissertations

Doctorate, Nöroblastom'da N-myc Onkogen Amplification ve klinik önemi, Istanbul University, Sağlık Bilimleri Enstitüsü, 1994

Academic Titles / Tasks

Professor, Acibadem Mehmet Ali Aydinlar University, Vocational School Of Health Services, 2016 - Continues
Professor, Istanbul University, Deneysel Tıp Araştırma Enstitüsü, Genetik Anabilim Dalı, 2003 - Continues
Professor, Erasmus Universiteit Rotterdam, Erasmus Medical Faculty, Erasmus Medical Center, 2006 - 2007
Research Assistant, University of Tennessee, Memphis, St. Jude Children's Rerearch Hospital, Genetics Department, 1998 - 2000

Academic and Administrative Experience

Head of Department, Acibadem Mehmet Ali Aydinlar University, Graduate School Of Health Sciences, Translasyonel Tıp Anabilim Dalı, 2022 - Continues
Head of Department, Acibadem Mehmet Ali Aydinlar University, Graduate School Of Health Sciences, Department Of Genomic Studies, 2021 - Continues
University Executive Board Member, Acibadem Mehmet Ali Aydinlar University, Administrative Departments, 2018 - Continues
Director of the Center, Acibadem Mehmet Ali Aydinlar University, Additional Departments, 2017 - Continues
Head of Department, Acibadem Mehmet Ali Aydinlar University, School Of Medicine, Department Of Medical Sciences, 2016 - Continues
Director of The Institution, Acibadem Mehmet Ali Aydinlar University, 2016 - 2022
Istanbul University, 2010 - 2016

Courses

Thesis planning and management, Doctorate, 2022 - 2023
Diabetes Genetics, Undergraduate, 2022 - 2023
Neurogenetics, Undergraduate, 2022 - 2023
GIS Diseases genetics, Undergraduate, 2021 - 2022
Project planning and management, Doctorate, 2021 - 2022
Hematological Malignancies, Undergraduate, 2021 - 2022

Advising Theses

Özbek U., Yaygın değişken immün yetersizlik (CVID) hastalarında CD19, ICOS, TACI, BAFF-R gen mutasyonlarının araştırılması, Postgraduate, S.SISKO(Student), 2016
Özbek U., Çocukluk çağı nüks akut lösemi hastalarında tüm genom analizleri, Doctorate, Y.ERBİLGİN(Student), 2015
Özbek U., Ailesel geçiş gösteren idiyopatik epilepsilerin genetik altyapısının aydınlatılması, Doctorate, F.NUR(Student), 2014
Özbek U., Stem-loop RT-polimeraz zincir reaksiyonu (PZR) yöntemi ile mikro RNA (miRNA) gen anlatım farklılıklarının tespiti, Postgraduate, N.MAVİ(Student), 2013
Özbek U., T-hücreli akut lenfoblastik lösemide genom boyu anlatım analizleri, Doctorate, Ö.HATIRNAZ(Student), 2012
Özbek U., T hücreli akut lenfoblastik lösemili hastalarda mikroRNA (miRNA) anlatımının tespiti, Postgraduate, C.ÖZTUNÇ(Student), 2012
Özbek U., İdiyopatik jeneralize epilepsili ailelerde lokus ve gen analizi, Postgraduate, İ.KARACAN(Student), 2012
Özbek U., Mezyal temporal lob epilepsisinde apoptoza yol açan genlerin ekspresyonlarının analizi, Postgraduate, E.YÜCESAN(Student), 2011
Özbek U., B hücreli akut lenfoblastik lösemilerde PAX5 geninin araştırılması, Postgraduate, S.ÖZDEMİRLİ(Student), 2010
Özbek U., Temporal lob epilepsi patogenezinde enflamasyonla ilgili genlerin ekspresyonlarının araştırılması, Postgraduate, Ö.ÖZDEMİR(Student), 2010
Özbek U., PINK1 Ve SWAP70 genlerinin klonlanmasıyla oluşturulan proteinlerin behçet hastalarında ELISA yöntemi ile taranması, Postgraduate, E.UĞUREL(Student), 2009
Özbek U., Temporal lob epileptogeneziyle ilişkili genlerin ekspresyon analizi, Doctorate, N.BEBEK(Student), 2009
Özbek U., Çocukluk çağı akut myeloid lösemi hastalarında kromozomal değişiklikler ve FLT3 geni mutasyonlarının araştırılması, Postgraduate, E.MEHMET(Student), 2008
Özbek U., T-ALL hastalarında NOTCH1 mutasyonunun araştırılması, Postgraduate, Y.ERBİLGİN(Student), 2008
Özbek U., Akut myeloid lösemi etiyolojisinde MN1 geninin rolü, Doctorate, S.SIRMA(Student), 2007
Özbek U., Behçet hastalığında rol alan yeni immunogenetik antijenlerin SEREX metodu ile belirlenmesi, Doctorate, B.VURAL(Student), 2005
Özbek U., CYP1A1, CYP2D6, CYP2E1, GSTT1 ve GSTM1 gen polimorfizmlerinin akut lösemi etiyolojisindeki rollerinin saptanması, Doctorate, M.AYDIN(Student), 2003
Özbek U., Sitokin gen polimorfizmlerinin hematopoetik kök hücre nakli sonrası graft-versus-host hastalığı ile ilişkisi, Postgraduate, N.GÜRSES(Student), 2003
Özbek U., Çocukluk çağı obesitesinde glukokortikoid reseptör gen polimorfizminin bazı somatik ve biyokimyasal parametrelerle ilişkisi, Postgraduate, N.ŞEN(Student), 2002
Özbek U., Medüller tiroid karsinomlu olgularda RET (REarranged during transfection) proto-onkogen mutasyonları, Postgraduate, N.BİÇER(Student), 1999
Özbek U., Çocukluk çağı akut lenfoblastik lösemilerinde TEL-AML1 (Translocation Ets Leukemia-Acute Myeloid Leukemia 1) füzyon geni tespiti ve prognostik önemi, Postgraduate, S.SIRMA(Student), 1999

Published journal articles indexed by SCI, SSCI, and AHCI

I. **Impact of TP53 gene variants on prognosis and survival of childhood acute lymphoblastic leukemia.**
Firtina S., Erbilgin Y., Hatirnaz Ng Ö., Karaman S., Karakas Z., Celkan T. T., Gelen S. A., Yildirmak Y., Ozbek U., Sayitoglu M.
Scandinavian journal of clinical and laboratory investigation, vol.83, no.3, pp.187-193, 2023 (SCI-Expanded)

II. **Perception and management of cancer predisposition in pediatric cancer centers: A European-wide questionnaire-based survey.**
Lazic J., Haas O. A., Özbek U., Ripperger T., Byrjalsen A., Te Kronnie G., Sayitoğlu M., Ng Ö., Agaoglu N. B., Erbilgin Y., et al.
Pediatric blood & cancer, vol.70, no.5, 2023 (SCI-Expanded)

III. **Preparation of Cell-Loaded Microbeads as Stable and Injectable Delivery Platforms for Tissue Engineering.**
Karaca M. A., Kançağı D., Ozbek U., Ovali E., Gök Ö.
Biomimetics (Basel, Switzerland), vol.8, no.2, 2023 (SCI-Expanded)

IV. **Obstacles and expectations of rare disease patients and their families in Turkiye: ISTisNA project survey results**
Hatirnaz Ng Ö., Sahin I., Erbilgin Y., Ozdemir O., Yucesan E., Erturk N., Yemenici M., Akgun Dogan Ö., Ugur Iseri S. A., Satman I., et al.
FRONTIERS IN PUBLIC HEALTH, vol.10, 2023 (SCI-Expanded)

V. **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., ÖZBEK U., UĞUR İŞERİ S. A.
Genes and Genomics, vol.45, no.1, pp.13-21, 2023 (SCI-Expanded)

VI. **Undiagnosed diseases: Needs and opportunities in 20 countries participating in the Undiagnosed Diseases Network International**
Taruscio D., Salvatore M., Lumaka A., Carta C., Cellai L. L., Ferrari G., Sciascia S., Groft S., ALANAY Y., Azam M., et al.
FRONTIERS IN PUBLIC HEALTH, vol.11, 2023 (SCI-Expanded)

VII. **Optimizing the Personalized Care for the Management of Rectal Cancer: A Consensus Statement**
Aytaç E., Özer L., Baca B., Balık E., Kapran Y., Taşkın O. C., Oyan Uluç B., Abacıoğlu M. U., Gönenç M., Bölükbaşı Y., et al.
TURKISH JOURNAL OF GASTROENTEROLOGY, vol.33, no.8, pp.627-663, 2022 (SCI-Expanded)

VIII. **Determining T and B Cell development by TREC/KREC analysis in primary immunodeficiency patients and healthy controls**
Senturk G., Ng Y. Y., Eltan S. B., Baser D., Ogulur I., Altindirek D., Firtina S., Yilmaz H., Kocamis B., Kiykim A., et al.
SCANDINAVIAN JOURNAL OF IMMUNOLOGY, vol.95, 2022 (SCI-Expanded)

IX. **Primary antibody deficiencies in Turkey: molecular and clinical aspects**
Firtina S., Ng Y. Y., HATIRNAZ NG Ö., Kiykim A., Ozek E. Y., Kara M., AYDINER E., Nepesov S., Camcioglu Y., Sayar E. H., et al.
IMMUNOLOGIC RESEARCH, vol.70, no.1, pp.44-55, 2022 (SCI-Expanded)

X. **Mutational landscape of SARS-CoV-2 genome in Turkey and impact of mutations on spike protein structure.**
Hatirnaz N., Akyoney S., Sahin I., Soykam H., Bayram A., Ozdemir Ö., Kancagi D., Sir K., Yurtsever B., Kocagoz A., et al.
PloS one, vol.16, no.12, 2021 (SCI-Expanded)

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

XII. **Prognostic evidence of LEF1 isoforms in childhood acute lymphoblastic leukemia.**
ERBİLGİN Y., Ng Ö., Can I., Firtina S., Kucukcankurt F., KARAMAN S., KARAKAŞ Z., Celkan T. T., ZENGİN E., AYLAN GELEN S., et al.
International journal of laboratory hematology, vol.43, no.5, pp.1093-1103, 2021 (SCI-Expanded)

XIII. **Bağlantı ve Tüm Ekzom Dizileme Analizlerinin Birlikte Değerlendirilmesiyle CIC Geninin İzole Distoni**

Adayı Olarak Belirlenmesi
Salman B., YÜCESAN E., SAMANCI B., BİLGİÇ B., HANAĞASI H. A., GÜRVİT İ. H., ÖZBEK U., UĞUR İŞERİ S. A.
JOURNAL OF ISTANBUL FACULTY OF MEDICINE-ISTANBUL TIP FAKULTESI DERGISI, vol.84, pp.457-463, 2021 (SCI-Expanded)

XIV. **Gamma-irradiated SARS-CoV-2 vaccine candidate, OZG-38.61.3, confers protection from SARS-CoV-2 challenge in human ACEII-transgenic mice.**
Turan R. D., Tastan C., Kancagi D. D., Yurtsever B., Karakus G. S., Ozer S., Abanuz S., Cakirsoy D., TÜMENTEMUR G., Demir S., et al.
Scientific reports, vol.11, no.1, pp.15799, 2021 (SCI-Expanded)

XV. **Preclinical efficacy and safety analysis of gamma-irradiated inactivated SARS-CoV-2 vaccine candidates**
Sir Karakus G., Tastan C., Dilek Kancagi D., Yurtsever B., Tumentemur G., Demir S., Turan R. D., Abanuz S., Cakirsoy D., Seyis U., et al.
SCIENTIFIC REPORTS, vol.11, no.1, 2021 (SCI-Expanded)

XVI. **Mutational landscape of severe combined immunodeficiency patients from Turkey**
Çekiç Ş., Ng Y. Y., Ng Ö., Kıykım A., Özen A. O., Özen A. O., Özen A. O., Özen A. O., Özen A. O., Torun S. H., et al.
INTERNATIONAL JOURNAL OF IMMUNOGENETICS, vol.47, no.6, pp.529-538, 2020 (SCI-Expanded)

XVII. **Copy-number variations in adult patients with chronic immune thrombocytopenia.**
YÜCESAN E., Ng Ö., Yalniz F. F., Yilmaz H., SALİHOĞLU A., Sudutan T., Eskazan A. E., Ongoren S., Baslar Z., Soysal T., et al.
Expert review of hematology, vol.13, no.11, pp.1277-1287, 2020 (SCI-Expanded)

XVIII. **Hydrogen Sulphide and Nitric Oxide Cooperate in Cardioprotection Against Ischemia/Reperfusion Injury in Isolated Rat Heart.**
ÜSTÜNOVA S., TAKIR S., YILMAZER N., Bulut H., ALTINDİREK D., HATIRNAZ NG Ö., Tansel C. D., Dogan B. S. U., ÖZBEK U., ARMUTAK E. İ., et al.
In vivo (Athens, Greece), vol.34, no.5, pp.2507-2516, 2020 (SCI-Expanded)

XIX. **The Impact and Prognostic Significance of Chronic Lymphocytic Leukemia Upregulated 1 (CLLU1) Gene Expression in Patients with Chronic Lymphocytic Leukemia: A Single Center Experience**
Sevinc M., Karabulut A., Eskazan A. E., Tatonyan S. C., ÖZBEK U., Soysal T.
LABORATORY MEDICINE, vol.51, no.3, pp.259-264, 2020 (SCI-Expanded)

XX. **PTEN and AKT1 Variations in Childhood T-Cell Acute Lymphoblastic Leukemia**
Kucukcankurt F., Erbilgin Y., Firtina S., Hatirnaz Ö., Karakas Z., Celkan T., Unuvar A., Ozbek U., Sayitoglu M.
TURKISH JOURNAL OF HEMATOLOGY, vol.37, no.2, pp.98-103, 2020 (SCI-Expanded)

XXI. **Association of genetic variants with colorectal cancer in the extended MENA region: A systematic review.**
Sidenna M., Bux R., Fadl T., Ozbek U., Zayed H.
Current molecular medicine, 2019 (SCI-Expanded)

XXII. **Homozygous c.130-131 ins A (pW44X) mutation in the HAX1 gene as the most common cause of congenital neutropenia in Turkey: Report from the Turkish Severe Congenital Neutropenia Registry.**
YILMAZ KARAPINAR D., PATIROĞLU T., Metin A., Caliskan U., CELKAN T. T., YILMAZ B., KARAKAŞ Z., Karapinar T. H., Akinci B., Ozkinay F., et al.
Pediatric blood & cancer, vol.66, no.10, 2019 (SCI-Expanded)

XXIII. **Prognostic gene alterations and clonal changes in childhood B-ALL.**
ERBİLGİN Y., Firtina S., Mercan S., Ng Ö., KARAMAN S., Tasar O., KARAKAŞ Z., Celkan T. T., Zenging E., Sarperg N., et al.
Leukemia research, vol.83, pp.106159, 2019 (SCI-Expanded)

XXIV. **The Outcomes of Chronic Myeloid Leukemia Patients With Molecular Warning Responses During Imatinib Treatment According to the European LeukemiaNet 2013 Recommendations**
Soysal T., Eskazan A. E., Serin I., Sadri S., Keskin D., Yurttas N. O., Berk S., Ozunal I. E., SALİHOĞLU A., AR M. C., et al.
CLINICAL LYMPHOMA MYELOMA & LEUKEMIA, vol.19, no.7, 2019 (SCI-Expanded)

XXV. **Dealing with the gray zones in the management of gastric cancer: The consensus statement of the**

Istanbul Group
AYTAÇ E., Aslan F., ÇİÇEK B., Erdamar S., Gurses B., GÜVEN K., Falay O., KARAHASANOĞLU T., Selcukbiricik F., Selek U., et al.
TURKISH JOURNAL OF GASTROENTEROLOGY, no.7, pp.584-598, 2019 (SCI-Expanded)

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

XXVII. **No evidence for a BRD2 promoter hypermethylation inblood leukocytes of Europeans with juvenile myoclonic epilepsy**
Schulz H., Ruppert A., Zara F., Madia F., Iacomino M., Vari M. S., Balagura G., Minetti C., Striano P., Blanche A., et al.
EPILEPSIA, vol.60, no.5, 2019 (SCI-Expanded)

XXVIII. **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., ARGÜDEN Y., ÇIRAKOĞLU A., ÇAKMUR R., DÖNMEZ ÇOLAKOĞLU B., Hacihanefioglu S., ÖZBEK U., Yapici Z., et al.
NEUROMOLECULAR MEDICINE, vol.21, no.1, pp.54-59, 2019 (SCI-Expanded)

XXIX. **Identification of epilepsy related pathways using genome-wide DNA methylation measures: A trio-based approach**
ÖZDEMİR Ö., Egemen E., UĞUR İŞERİ S. A., SEZERMAN O. U., BEBEK N., BAYKAL B., ÖZBEK U.
PLOS ONE, vol.14, no.2, 2019 (SCI-Expanded)

XXX. **; The Role of the Local Bone Marrow Renin-Angiotensin System in Multiple Myeloma**
Sakai B., SAYİTOĞLU M., İSTEMİHAN Z., KARAN M. A., ERTEN S. N., Dogan O., ÖZBEK U., GENÇ N. S., Tascioglu C., BEŞIŞIK S.
TURKISH JOURNAL OF HEMATOLOGY, vol.36, no.3, pp.178-185, 2019 (SCI-Expanded)

XXXI. **Deep sequencing of BCR-ABL1 kinase domain mutations in chronic myeloid leukemia patients with resistance to tyrosine kinase inhibitors.**
ERBİLGİN Y., Eskazan A. E., HATIRNAZ NG Ö., SALİHOĞLU A., ELVERDİ T., Firtina S., Tasar O., Mercan S., Sisko S., Khodzhaev K., et al.
Leukemia & lymphoma, vol.60, no.1, pp.200-207, 2019 (SCI-Expanded)

XXXII. **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. U., YAPICI Z.
NEUROLOGIA I NEUROCHIRURGIA POLSKA, vol.53, no.6, pp.476-483, 2019 (SCI-Expanded)

XXXIII. **Frontline nilotinib treatment in Turkish patients with Philadelphia chromosome-positive chronic Myeloid Leukemia in chronic phase: updated results with 2 years of follow-up**
SAYDAM G., HAZNEDAROĞLU İ. C., KAYNAR L., Yavuz A., ALİ R., GÜVENÇ B., AKAY O. M., Baslar Z., ÖZBEK U., SÖNMEZ M., et al.
HEMATOLOGY, vol.23, no.10, pp.771-777, 2018 (SCI-Expanded)

XXXIV. **Investigation of SLC2A1 gene variants in genetic generalized epilepsy patients with eyelid myoclonia**
Altiokka-Uzun G., ÖZDEMİR Ö., Ugur-Iseri S., BEBEK N., Gurses C., ÖZBEK U., BAYKAL B.
EPILEPTIC DISORDERS, vol.20, no.5, pp.396-400, 2018 (SCI-Expanded)

XXXV. **Rare coding variants in genes encoding GABA(A) receptors in genetic generalised epilepsies: an exome-based case-control study**
May P., Girard S., Harrer M., Bobbili D. R., Schubert J., Wolking S., Becker F., Lachance-Touchette P., Meloche C., Gravel M., et al.
LANCET NEUROLOGY, vol.17, no.8, pp.699-708, 2018 (SCI-Expanded)

XXXVI. **Clinical phenotype of hereditary spastic paraplegia due to KIF1C gene mutations across life span**
Yucel-Yilmaz D., YÜCESAN E., YALNIZOĞLU D., Oguz K. K., Sagiroglu M. S., ÖZBEK U., SERDAROĞLU E., BİLGİÇ B., Erdem S., UĞUR İŞERİ S. A., et al.

BRAIN & DEVELOPMENT, vol.40, no.6, pp.458-464, 2018 (SCI-Expanded)

XXXVII. **Association of Pro-apoptotic Bad Gene Expression Changes with Benign Thyroid Nodules**
GÜL N., Temel B., Ustek D., SIRMA EKMEKCİ S., Kapran Y., TUNCA F., ŞENYÜREK Y., ÖZBEK U., Alagol F.
IN VIVO, vol.32, no.3, pp.555-559, 2018 (SCI-Expanded)

XXXVIII. **Outcomes with frontline nilotinib treatment in Turkish patients with newly diagnosed Philadelphia chromosome-positive chronic myeloid leukemia in chronic phase.**
Saydam G., Haznedaroglu I., Kaynar L., Yavuz A., Ali R., Guvenc B., Akay O., Baslar Z., Ozbek U., Sonmez M., et al.
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XXXIX. **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., ELVERDİ T., Salihoglu A., ERBİLGİN Y., et al.
HEMATOLOGY, vol.23, no.4, pp.212-220, 2018 (SCI-Expanded)

XL. **Outcomes of Chronic Myeloid Leukemia Patients With Early Molecular Response at 3 and 6 Months: A Comparative Analysis of Generic Imatinib and Glivec**
Eskazan A. E., Sadri S., Keskin D., Ayer M., Kantarcioglu B., Demirel N., Aydin D., Aydinli F., Yokus O., Ozunal I. E., et al.
CLINICAL LYMPHOMA MYELOMA & LEUKEMIA, vol.17, no.12, pp.804-811, 2017 (SCI-Expanded)

XLI. **SYNE1 related cerebellar ataxia presents with variable phenotypes in a consanguineous family from Turkey**
YÜCESAN E., UĞUR İŞERİ S. A., BİLGİÇ B., Gormez Z., GÜNGÖR B., Sarac A., ÖZDEMİR Ö., Sagiroglu M., GÜRVİT İ. H., HANAĞASI H. A., et al.
NEUROLOGICAL SCIENCES, vol.38, no.12, pp.2203-2207, 2017 (SCI-Expanded)

XLII. **A novel pathogenic frameshift variant of CD3E gene in two T-B+ NK+ SCID patients from Turkey.**
Firtina S., Ng Y. Y., Ng Ö., Nepesov S., Yesilbas O., Kilercik M., Burtecene N., Cinar S., Camcioglu Y., ÖZBEK U., et al.
Immunogenetics, vol.69, no.10, pp.653-659, 2017 (SCI-Expanded)

XLIII. **Clinical and genetic features of PKAN patients in a tertiary centre in Turkey.**
Akcakaya N. H., Iseri S. U., Bilir B., Battaloglu E., Tekturk P., GÜLTEKİN M., Akar G., Yigiter R., Hanagasi H., Alp R., et al.
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XLIV. **Dysregulation of the DKK1 gene in pediatric B-cell acute lymphoblastic leukemia**
Firtina S., Hatirnaz Ng Ö., Erbilgin Y., Ozbek U., Sayitoglu M.
TURKISH JOURNAL OF MEDICAL SCIENCES, vol.47, no.1, pp.357-363, 2017 (SCI-Expanded)

XLV. **Aberrant Hypermethylation of APC Tumor Supressor Gene in Acute Leukemia Patients**
Hatirnaz O. N. G., Firtina S., Erbilgin Y., Özbek U., Sayitoglu M.
UHOD-ULUSLARARASI HEMATOLOJI-ONKOLOJI DERGISI, vol.27, pp.1-7, 2017 (SCI-Expanded)

XLVI. **Outcomes with frontline nilotinib treatment in Turkish patients with newly diagnosed Philadelphia chromosome-positive chronic myeloid leukemia in chronic phase**
SAYDAM G., HAZNEDAROĞLU İ. C., KAYNAR L., Yavuz A., ALİ R., GÜVENÇ B., AKAY O. M., Baslar Z., ÖZBEK U., SÖNMEZ M., et al.
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XLVII. **A novel gene mutation in PANK2 in a patient with severe jaw-opening dystonia.**
Yapici Z., Akcakaya N. H., Tekturk P., Iseri S. A. U., Ozbek U.
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XLVIII. **Imatinib reduces bone marrow fibrosis and overwhelms the adverse prognostic impact of reticulin formation in patients with chronic myeloid leukaemia**
Simsek E. T., Eskazan A. E., Cengiz M., AR M. C., Ekizoglu S., Salihoglu A., Gulturk E., ELVERDİ T., Aydin S. O., DEMİRÖZ A. S., et al.
JOURNAL OF CLINICAL PATHOLOGY, vol.69, no.9, pp.810-816, 2016 (SCI-Expanded)

XLIX. **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., YÜCESAN E., TUNCER KILINÇ F. N., ÖZDEMİR Ö., BEBEK N., ÖZBEK U., UĞUR İŞERİ S. A.,

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L. **The frequency of C609T polymorphism in the NQO1 gene and its relation to cytogenetic abnormalities in patients with myelodysplastic syndrome**
Bagatir G., Sirma S., PALANDUZ Ş., ÖZTÜRK Ş., ÇEFLE K., ÖZBEK U., YENEREL M. N., NALÇACI M.
CELLULAR AND MOLECULAR BIOLOGY, vol.62, no.7, pp.61-65, 2016 (SCI-Expanded)

LI. **Investigation of the possible association of NEDD4-2 (NEDD4L) gene with idiopathic photosensitive epilepsy**
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Activities in Scientific Journals

FRONTIERS IN GENETICS, Evaluation Committee Member, 2019 - Continues

Memberships / Tasks in Scientific Organizations

Rare Disease International-first workshop on Strengthening the National Healthcare Systems, Member of Science Committee, 2022 - Continues, United States Of America
Tıbbi Genetik Derneği Nadir Hastalıklar Kurulu, Chairman of the Scientific Committee, 2021 - Continues, Turkey
International Cerebral Palsy Genomic Consortium, Executive Board Member, 2020 - Continues, Australia
Türkiye Spastik Çocuklar Vakfı, Member of Science Committee, 2017 - Continues, Turkey
UDNI-Undiagnosed Disease Network International, Member, 2016 - Continues, United States Of America
TÜSEB-TÜHKE, Member of Advisory Board, 2016 - Continues, Turkey
ORPHANET, Board Member, 2009 - Continues, France
BFM Biology and Diagnosis Committee, Principal Member, 2001 - Continues, Germany
Europeand Society of Human Genetics, Member, 1989 - Continues, Belgium

Scientific Refereeing

TUBITAK Project, 1004 - Mükemmeliyet Merkezi Destek Programı FAZ I, iBG, Turkey, October 2022
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Scientific Consultations

Cumhurbaşkanlığı Strateji ve Bütçe Başkanlığı, Scientific Consultancy, İstanbul Üniversitesi, Deneysel Tıp Araştırma Enstitüsü, Genetik Anabilim Dalı, Turkey, 2022 - 2023

Tasks In Event Organizations

Özbek U., UDNI meeting-November 7-8, 2022 in Vienna, Austria, Scientific Congress, Austria, Kasım 2022
Özbek U., Childhood Cancer: Causes and Epidemiology – A Course by SIOP-Europe and COST-LEGEND , Workshop Organization, Brussels, Belgium, Mayıs 2022
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Metrics

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Congress and Symposium Activities

6. Uluslararası ve 24. Ulusal Halk Sağlığı Kongresi, Invited Speaker, Antalya, Turkey, 2022
15. Uluslararası Katılımlı Ulusal Tıbbi Genetik Kongresi, Session Moderator, Muğla, Turkey, 2022
Undiagnosed Disease Network International Conferance , Attendee, Vienna, Austria, 2022
Türk Pediatrik Hematoloji Derneği Lösemi Çalıştayı, Panelists, Bursa, Turkey, 2022
"Türk Pediatrik Hematoloji Derneği Lösemi Çalıştayı, Invited Speaker, Bursa, Turkey, 2022
7. Erciyes Uluslararası Tıp Tıbbi Genetik Kongresi, Invited Speaker, Kayseri, Turkey, 2022
12. OHSAD kurultayı, Invited Speaker, Antalya, Turkey, 2022
Tıbbi Genetik Derneği Nadir Hastalıklar Günü sunumu, Invited Speaker, İstanbul, Turkey, 2022