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Personal Information

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Education Information

Doctorate, Hacettepe University, Fen Bilimleri Enstitüsü, Biyoloji/Moleküler Biyoloji, Turkey 2004 - 2010
Postgraduate, Hacettepe University, Fen Bilimleri Enstitüsü, Biyoloji/Moleküler Biyoloji, Turkey 2001 - 2004
Undergraduate, Hacettepe University, Fen Fakültesi, Biyoloji, Turkey 1997 - 2001

Foreign Languages

English, B2 Upper Intermediate

Dissertations

Doctorate, Kalıtsal Retina Dejenerasyonlarında DNA Mikroarray Yöntemiyle Yüksek Ölçekli Genom Taraması , Hacettepe Üniversitesi, Fen Fakültesi, Biyoloji Bölümü, 2010
Postgraduate, Yaşa Bağlı Maküler Dejenerasyonlu (AMD) Türk Hastalarda ABCR Geninde Sekans Değişikliklerinin Saptanması, Hacettepe Üniversitesi, Fen Bilimleri Enstitüsü, Moleküler Biyoloji (Yl) (Tezli), 2004

Research Areas

Medicine, Health Sciences, Internal Medicine Sciences, Child Health and Diseases, Life Sciences, Molecular Biology and Genetics, Genetic Disorders, Natural Sciences

Academic Titles / Tasks

Assistant Professor, Hacettepe University, Çocuk Sağlığı Enstitüsü, Pediatrik Temel Bilimler A.B.D., 2015 - Continues

Published journal articles indexed by SCI, SSCI, and AHCI

I. **The development and validation of a scale on L2 writing teacher feedback literacy**
Lee I., Karaca M., Inan S.
Assessing Writing, vol.57, 2023 (SSCI)
II. **An investigation into L2 writing teacher beliefs and their possible sources**
Karaca M., Uysal H. H.
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III. **Researching and Teaching Second Language Writing in the Digital Age**

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IV. **Biallelic mutations in ELFN1 gene associated with developmental and epileptic encephalopathy and joint laxity**
DURSUN A., YALNIZOĞLU D., Yilmaz D. Y., Oguz K. K., GÜLBAKAN B., KOŞUKCU C., AKAR H. T., Kahraman A. B., Acar N. V., GÜNBEY C., et al.
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V. **DNACJ12 deficiency in patients with unexplained hyperphenylalaninemia: two new patients and a novel variant**
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VI. **Clinical and molecular characteristics of carnitine-acylcarnitine translocase deficiency with c.270delC and a novel c.408C>A variant**
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VIII. **Clinical highlights of a very rare phospolipid remodeling disease due to MBOAT7 gene defect**
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AMERICAN JOURNAL OF MEDICAL GENETICS PART B-NEUROPSYCHIATRIC GENETICS, 2019 (SCI-Expanded)

IX. **Expanding the phenotype of phospholipid remodelling disease due to MBOAT7 gene defect**
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X. **Clinical phenotype of hereditary spastic paraplegia due to KIF1C gene mutations across life span**
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XII. **Genotypic-phenotypic features and enzyme replacement therapy outcome in patients with mucopolysaccharidosis VI from Turkey**
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XIII. **A patient with mitochondrial disorder due to a novel mutation in MRPS22**
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XIV. **A probable new syndrome with the storage disease phenotype caused by the VPS33A gene mutation**
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XV. **Evaluation and identification of IDUA gene mutations in Turkish patients with mucopolysaccharidosis type I**
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XVI. **Sapropterin dihydrochloride treatment in Turkish hyperphenylalaninemic patients under age four**
ÖZLEM U., GÖKMEN ÖZEL H., COŞKUN T., ÖZGÜL R. K., YÜCEL YILMAZ D., BURCU H., TOKATLI A., DURSUN A., SİVRİ H. S.

Turkish Journal Of Pediatrics, pp.213-218, 2015 (SCI-Expanded)

XVII. **Detection of biotinidase gene mutations in Turkish patients ascertained by newborn and family screening**
KARACA M., ÖZGÜL R. K., ÜNAL O., Yucel-Yilmaz D., KILIÇ M., Hismi B., TOKATLI A., COŞKUN T., DURSUN A., SİVRİ H. S.
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XVIII. **Two Turkish siblings with MEGDEL syndrome due to novel SERAC1 genemutation.**
ÜNAL Ö., ÖZGÜL R. K., YÜCEL YILMAZ D., YALNIZOĞLU D., TOKATLI A., SİVRİ H. S., HİŞMİ B., COŞKUN T., DURSUN A.
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XIX. **Genome-Wide Homozygosity Mapping in Families with Leber Congenital Amaurosis Identifies Mutations in AIPL1 and RDH12 Genes**
Yucel-Yilmaz D., TARLAN B., KIRATLI H., ÖZGÜL R. K.
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XX. **Phenotypic and genotypic spectrum of Turkish patients with isovaleric acidemia**
ÖZGÜL R. K., Karaca M., Kilic M., KUCUK O., YUCEL-YILMAZ D., UNAL O., HISMI B., Aliefendioglu D., SIVRI S., TOKATLI A., et al.
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XXI. **Dursun Syndrome Due to G6PC3 Gene Defect has a Fluctuating Pattern in All Blood Cell Lines**
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Yucel D., YILMAZ M., DURUKAN A. H., ÖZGÜL R. K.
OPHTHALMIC GENETICS, vol.33, no.3, pp.144-149, 2012 (SCI-Expanded)

XXVI. **Comparative analysis of genetic diversity in Turkish durum wheat cultivars using RAPD and ISSR markers**
Karaca M., İZBIRAK A.
JOURNAL OF FOOD AGRICULTURE & ENVIRONMENT, vol.6, pp.219-225, 2008 (SCI-Expanded)

Articles Published in Other Journals

I. **HOMOZYGOUS GNAL MUTATION ASSOCIATED WITH FAMILIAL CHILDHOOD-ONSET GENERALIZED DYSTONIA**
MASUHO I., FANG M., GENG C., ZHANG J., JİANG H., ÖZGÜL R. K., Yilmaz D. Y., YALNIZOĞLU D., YÜKSEL D., YARROW A., et al.
NEUROLOGY-GENETICS, vol.2, no.3, 2016 (ESCI)

II. **A Case of Glutaric Aciduria Type I with a Novel Mutation**
Unal N. U., KOR D., YÜCEL YILMAZ D., GÜL MERT G., Mungan N. O.

CUKUROVA MEDICAL JOURNAL, vol.38, no.4, pp.809-812, 2013 (ESCI)

Books & Book Chapters

I. **Identification of mutations and evaluation of cardiomyopathy in Turkish patients with primary carnitine deficiency.**
KILIÇ M., ÖZGÜL R. K., COŞKUN T., YÜCEL YILMAZ D., KARACA M., SİVRİ H. S., TOKATLI A., ŞAHİN M., KARAGÖZ T., DURSUN A.
in: JIMD Reports Case and Research Reports 2011 3, , Editor, SPRINGER, 2011

Refereed Congress / Symposium Publications in Proceedings

I. **Dirençli Hipoglisemi Hipertrofik Kardiyomiyopati ve Ensefalopatili Bir Hastada Ekzom Dizileme ile Mitokondriyal TSFM Gen Defekti**
DURSUN A., ÖMER FARUK G., MELİS P., YÜCEL YILMAZ D., TOPÇU M., YALNIZOĞLU D., YİĞİT Ş., ORHAN D., MAHMUT S., ÖZGÜL R. K.
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Supported Projects

DURSUN A., ÖZGÜL R. K., YÜCEL YILMAZ D., Project Supported by Higher Education Institutions, MBOAT7 Gen Defektinin Protein İfadesinin Araştırılması, 2019 - 2022
DURSUN A., DÖKMECİ S., YÜCEL YILMAZ D., TUBITAK Project, Yeni Tanımlanan Metabolik/Nörometabolik Hastalıklarda Otofaji Mekanizmasının Araştırılması, 2018 - 2021
DURSUN A., YALNIZOĞLU D., YÜCEL YILMAZ D., ÖZGÜL R. K., Project Supported by Higher Education Institutions, TANI KONULAMAYAN METABOLİK/NÖROMETABOLİK HASTALIKLARDA VEZİKÜLER TRAFİK BOZUKLUKLARININ ARAŞTIRILMASI, 2017 - 2021
ÖZSOY E. D., AKARSU A. N., Gözüböyük M., YILMAZ M., YÜCEL YILMAZ D., Project Supported by Higher Education Institutions, Gelişim genetiği ve biyolojisi açısından önemli olan aristaless ve lim homeobox 1 genlerine ait gen etkileşim ağlarının Drosophila melanogaster Genom Referans Paneli soyları üzerinden epistatik genomik haritalama yöntemi ile saptanması, 2017 - 2020
Yücel Yılmaz D., Gülbakan B., TUBITAK Project, MBOAT7 GEN MUTASYONU OLAN HASTALARDA AYIRICI TANI İÇİN KÜTLE SPEKTROMETRİ TEMELLİ METABOLOMİK PROFİLLEME İLE BİYOBELİRTEÇ KEŞFİ, 2017 - 2020
YÜCEL YILMAZ D., Project Supported by Higher Education Institutions, NÖROMETABOLİK HASTALIK TANISI İLE İZLENEN BİR AİLEDE EKZOM VERİLERİNİN DEĞERLENDİRİLMESİ VE SORUMLU GENİN TANIMLANMASI, 2017 - 2017
TAN Ç., ÖZBEK B., KARAPINAR Ö., TEZCAN F. İ., ÇAĞDAŞ AYVAZ D. N., YÜCEL YILMAZ D., Project Supported by Higher Education Institutions, MUHTEMEL ALPS TANISI ALAN HASTALARDA FAS GEN EKSPRESYONU TAYİNİ, 2016 - 2017
TAN Ç., ÇAĞDAŞ AYVAZ D. N., TEZCAN F. İ., YÜCEL YILMAZ D., ÖZBEK B., Project Supported by Higher Education Institutions, Primer Antikor Eksikliğine İlişkin Genetik Hastalıkların Yeni Nesil Dizileme Yöntemiyle Taranması, 2016 - 2017
YÜCEL YILMAZ D., Project Supported by Higher Education Institutions, Etilmalonik asidüri saptanmayan etilmalonik ensefalopati vakası, 2016 - 2016

Metrics

Publication: 45
Citation (WoS): 164
Citation (Scopus): 247

H-Index (WoS): 9
H-Index (Scopus): 10