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Personal Information

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Education Information

Doctorate, Hacettepe Üniversitesi, Fen Bilimleri Enstitüsü, Biyoloji/Moleküler Biyoloji, Turkey 2004 - 2010

Post Graduate, Hacettepe Üniversitesi, Fen Bilimleri Enstitüsü, Biyoloji/Moleküler Biyoloji, Turkey 2001 - 2004

Under Graduate, Hacettepe Üniversitesi, 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

Post Graduate, Yaşa Bağlı Maküler Dejenerasyonlu (AMD) Türk Hastalarda ABCR Geninde Sekans Değişikliklerinin Saptanması, Hacettepe Üniversitesi, Fen Bilimleri Enstitüsü, Moleküler Biyoloji (YI) (Tezli), 2004

Research Areas

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

Articles Published in Journals That Entered SCI, SSCI and AHCI Indexes

- I. **Glutaric aciduria type 1: Genetic and phenotypic spectrum in 53 patients**
BİLGİNER GÜRBÜZ B., YÜCEL YILMAZ D., COŞKUN T., TOKATLI A., DURSUN A., SİVRİ H. S.
EUROPEAN JOURNAL OF MEDICAL GENETICS, vol.63, no.11, 2020 (Journal Indexed in SCI)
- II. **A measure of possible sources of demotivation in L2 writing: A scale development and validation study**
Karaca M., Inan S.
ASSESSING WRITING, vol.43, pp.63-77, 2020 (Journal Indexed in SSCI)
- III. **Clinical highlights of a very rare phospholipid remodeling disease due to MBOAT7 gene defect**
DURSUN A., YALNIZOĞLU D., ÖZGÜL R. K. , Oguz K. K. , Yucel-Yilmaz D.
AMERICAN JOURNAL OF MEDICAL GENETICS PART B-NEUROPSYCHIATRIC GENETICS, 2019 (Journal Indexed in SCI)
- IV. **Expanding the phenotype of phospholipid remodelling disease due to MBOAT7 gene defect**
YALNIZOĞLU D., ÖZGÜL R. K. , Oguz K. K. , Ozer B., Yucel-Yilmaz D., Gurbuz B., Serdaroglu E., Erol I., Topcu M., DURSUN A.

JOURNAL OF INHERITED METABOLIC DISEASE, vol.42, no.2, pp.381-388, 2019 (Journal Indexed in SCI)

- V. **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. U. , et al.
BRAIN & DEVELOPMENT, vol.40, no.6, pp.458-464, 2018 (Journal Indexed in SCI)
- VI. **Five novel ALMS1 gene mutations in six patients with Alstrom syndrome**
Kilinc S., Yucel-Yilmaz D., Ardagil A., Apaydin S., Valverde D., ÖZGÜL R. K. , Guven A.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.31, no.6, pp.681-687, 2018 (Journal Indexed in SCI)
- VII. **Genotypic-phenotypic features and enzyme replacement therapy outcome in patients with mucopolysaccharidosis VI from Turkey**
Kilic M., DURSUN A., COŞKUN T., TOKATLI A., ÖZGÜL R. K. , YUCEL-YILMAZ D., Karaca M., Dogru D., ALEHAN D., KADAYIFÇILAR S., et al.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.173, no.11, pp.2954-2967, 2017 (Journal Indexed in SCI)
- VIII. **A patient with mitochondrial disorder due to a novel mutation in MRPS22**
Kilic M., Oguz K., Kilic E., YÜKSEL D., DEMİRCİ H., SAĞIROĞLU M. S. , Yucel-Yilmaz D., ÖZGÜL R. K.
METABOLIC BRAIN DISEASE, vol.32, no.5, pp.1389-1393, 2017 (Journal Indexed in SCI)
- IX. **A probable new syndrome with the storage disease phenotype caused by the VPS33A gene mutation**
DURSUN A., YALNIZOĞLU D., Gerdan O. F. , Yucel-Yilmaz D., Sagiroglu M. S. , YUKSEL B., GUCER S., Sivri S., ÖZGÜL R. K.
CLINICAL DYSMORPHOLOGY, vol.26, no.1, pp.1-12, 2017 (Journal Indexed in SCI)
- X. **Evaluation and identification of IDUA gene mutations in Turkish patients with mucopolysaccharidosis type I**
Atceken N., ÖZGÜL R. K. , Yilmaz D. Y. , TOKATLI A., COŞKUN T., SİVRİ H. S. , DURSUN A., Karaca M.
TURKISH JOURNAL OF MEDICAL SCIENCES, vol.46, no.2, pp.404-408, 2016 (Journal Indexed in SCI)
- XI. **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 (Journal Indexed in SCI Expanded)
- XII. **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.
EUROPEAN JOURNAL OF PEDIATRICS, vol.174, no.8, pp.1077-1084, 2015 (Journal Indexed in SCI)
- XIII. **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.
Turkish Journal Of Pediatrics, vol.57, pp.388-393, 2015 (Journal Indexed in SCI Expanded)
- XIV. **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.
DNA AND CELL BIOLOGY, vol.33, no.12, pp.876-883, 2014 (Journal Indexed in SCI)
- XV. **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., Aliefendioğlu D., SIVRI S., TOKATLI A., et al.
EUROPEAN JOURNAL OF MEDICAL GENETICS, vol.57, no.10, pp.596-601, 2014 (Journal Indexed in SCI)
- XVI. **Dursun Syndrome Due to G6PC3 Gene Defect has a Fluctuating Pattern in All Blood Cell Lines**
ÖZGÜL R. K. , YUCEL-YILMAZ D., DURSUN A.
JOURNAL OF CLINICAL IMMUNOLOGY, vol.34, no.3, pp.265-266, 2014 (Journal Indexed in SCI)
- XVII. **High prevalence of cerebral venous sinus thrombosis (CVST) as presentation of cystathionine beta-synthase deficiency in childhood: Molecular and clinical findings of Turkish probands**

- Karaca M., Hismi B., ÖZGÜL R. K., Karaca S., Yilmaz D. Y., COŞKUN T., SİVRİ H. S., TOKATLI A., DURSUN A.
GENE, vol.534, no.2, pp.197-203, 2014 (Journal Indexed in SCI)
- XVIII. **Galactosemia in the Turkish population with a high frequency of Q188R mutation and distribution of Duarte-1 and Duarte-2 variations**
Oezgul R. K., Guezel-Ozantuerk A., Duendar H., Yucel-Yilmaz D., COŞKUN T., Sivri S., Aydogdu S., Tokatli A., DURSUN A.
JOURNAL OF HUMAN GENETICS, vol.58, no.10, pp.675-678, 2013 (Journal Indexed in SCI)
- XIX. **Association of CFH Y402H Polymorphism with Both Forms of Advanced Age-Related Macular Degeneration in Turkish Patients**
Yucel D., YILMAZ M., DURUKAN A. H., ÖZGÜL R. K.
OPHTHALMIC GENETICS, vol.33, no.3, pp.144-149, 2012 (Journal Indexed in SCI)
- XX. **OCTN2 GENE MUTATIONS IN TURKISH PATIENTS WITH PRIMARY CARNITINE DEFICIENCY**
Yucel-Yilmaz D., Ersoy M., Candan S., Balci M., KILIÇ M., Gokcay G., DURSUN A., ÖZGÜL R. K.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.35, 2012 (Journal Indexed in SCI)
- XXI. **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 (Journal Indexed in SCI)

Articles Published in Other Journals

- I. **HOMOZYGOUS GNAL MUTATION ASSOCIATED WITH FAMILIAL CHILDHOOD-ONSET GENERALIZED DYSTONIA**
MASUHO I., FANG M., GENG C., ZHANG J., JIANG H., ÖZGÜL R. K., Yilmaz D. Y., YALNIZOĞLU D., YÜKSEL D., YARROW A., et al.
NEUROLOGY-GENETICS, vol.2, no.3, 2016 (Journal Indexed in 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 (Journal Indexed in 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. **Severe Motor Mental Retardation with Microcephaly and Hypomyelination due to PYCR2 Gene variant in a Large Family**
BİLGİNER GÜRBÜZ B., EROĞLU ERTUĞRUL N. G., KOŞUKCU C., YÜCEL YILMAZ D., ÖZGÜL R. K., YALNIZOĞLU D., DURSUN A.
Annual Symposium of the Society for the Study of Inborn Errors of Metabolism 2019 (SSIEM-2019), Rotterdam, Netherlands, 3 - 06 September 2019, vol.42, pp.1-479
- II. **Hyperphenylalaninemia due to novel JCDNA12 mutation**
SİVRİ H. S., ÇIKI K., YÜCEL YILMAZ D., GÜRSES CİLA H. E., ÖZGÜL R. K., TOKATLI A., COŞKUN T., DURSUN A.

SSIEM 2019: Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Rotterdam, Netherlands, 3 - 06 September 2019, vol.42, pp.324

- III. **The clinical, biochemical features, and mutational analyses in glutaric acid type 1 patients**
BİLGİNER GÜRBÜZ B., YILDIZ Y., GOKSOY E., YÜCEL YILMAZ D., DURSUN A., TOKATLI A., COŞKUN T., SİVRİ H. S.
International Congress of Inborn Errors of Metabolism - ICIEM 2017, 5 - 08 September 2017, vol.5
- IV. **pontocerebellar hypoplasia type 6 a case with neonatal seizures hypotonia and microcephaly diagnosed by exome sequencing**
DURSUN A., sedaroğlu e., ÖZGÜL R. K. , YÜCEL YILMAZ D., YALNIZOĞLU D.
SSIEM roma, 6 - 09 September 2016
- V. **ethymelanonic encephalopathy without etilmelanoc acitürria**
YÜCEL YILMAZ D., ÖZGÜL R. K. , PEKTAŞ E., SERDARDĞLU e., YALNIZOĞLU D., DURSUN A.
SSIEM ROMA, 6 - 09 September 2016
- VI. **FONKSİYONEL ÇALIŞMALAR Genetik uzmanlar için yeni nesil dizileme Kursu**
YÜCEL YILMAZ D.
3. Nörometabolik Dismorfoloji Sempozyumu, Turkey, 10 - 12 March 2016
- VII. **SNX14 Sorting Nexin 14 gene mutation causes a new syndromic form of cerebellar atrophy in a Turkish family**
ÖZGÜL R. K. , YÜCEL YILMAZ D., Ömer g., YALNIZOĞLU D., Mahmut s., DURSUN A.
SSIEM, 4 - 07 September 2015
- VIII. **Rhizomelic chondrodysplasia punctata type II a case diagnosed by whole exome sequencing**
DURSUN A., PEKTAŞ E., YÜCEL YILMAZ D., ÖZGÜL R. K.
SSIEM, 4 - 07 September 2015
- IX. **Mutation screening study in Turkish patients with L 2 hydroxyglutaric aciduria**
YÜCEL YILMAZ D., ÖZGÜL R. K. , Özlem u., COŞKUN T., SİVRİ H. S. , TOKATLI A., DURSUN A.
SSIEM, 4 - 07 September 2015
- X. **Splicing mutation in aminophospholipid transporter protein ATP8A2 in a Turkish family**
DURSUN A., YALNIZOĞLU D., YÜCEL YILMAZ D., serdaroğlu E., Unal Ö., görmez Z., Demirci H., Sağıroğlu M., ÖZGÜL R. K.
SSIEM, 4 - 06 September 2015
- XI. **Exome sequencing results in unknown genetic metabolic neurometabolic disorders**
KILIÇ M., ÖZGÜL R. K. , KILIÇ E., YÜCEL YILMAZ D., KAVAK P., YUCETURK B., DEMİRCİ H., SAĞIROĞLU M.
Annual Symposium of the Society for the Study of Inborn Errors of Metabolism (SSIEM), 1 - 04 September 2015
- XII. **A case of fucosidosis with a new mutation in FUCA1 gene**
PEKTAŞ E., YÜCEL YILMAZ D., ÖZGÜL R. K. , DURSUN A.
Annual Symposium of the Society for the Study of Inborn Errors of Metabolism (SSIEM), 1 - 04 September 2015
- XIII. **Galactosemia case with a novel mutation**
KILIÇ M., ZENCİROĞLU A., GÖKSUN E., YÜCEL YILMAZ D., ÖZGÜL R. K.
Annual Symposium of the Society for the Study of Inborn Errors of Metabolism (SSIEM), 1 - 04 September 2015
- XIV. **Vacuolar storage material in a family with juvenile parkinsonism and mutations in FBXO7**
ESRA S., ÖZGÜL R. K. , YALNIZOĞLU D., MADEO M., MALANDRİNİ A., KLEE E., Lİ Y., TN J., KARLI OĞUZ H. K. , YÜCEL YILMAZ D., et al.
Annual Symposium of the Society for the Study of Inborn Errors of Metabolism (SSIEM), 1 - 04 September 2015
- XV. **Hereditary spastic paraplegia with predominant cerebellar signs due to KIF1C mutation in two brothers**
ÖZGÜL R. K. , Esra s., YALNIZOĞLU D., YÜCEL YILMAZ D., TOPÇU M., ZZ G., Sağıroğlu M., DURSUN A.
SSIEM, 4 - 07 September 2015

Supported Projects

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

Citations

Total Citations (WOS):110

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