

Prof. Dr. ALİ DURSUN

Kişisel Bilgiler

E-posta: adursun@hacettepe.edu.tr

Web: <https://avesis.hacettepe.edu.tr/adursun>

Uluslararası Araştırmacı ID'leri

ORCID: 0000-0003-1104-9902

Yoksis Araştırmacı ID: 22512

Eğitim Bilgileri

Tıpta Uzmanlık, Hacettepe Üniversitesi, Tıp Fakültesi, Çocuk Sağlığı Hastalıkları Anabilim Dalı, Türkiye 1988 - 1992

Yaptığı Tezler

Doktora, Türk Akça ağacı şurubu hastalarında dallı zincirli Alpha - keto asit dehidrogenaz enzim kompleksi mutasyonlarının araştırılması, İstanbul Üniversitesi, Deneysel Tıp Araştırma Enstitüsü, Genetik Anabilim Dalı, 1998

Araştırma Alanları

Tıp, Sağlık Bilimleri, Dahili Tıp Bilimleri, Çocuk Sağlığı ve Hastalıkları, Pediatrik Endokrinoloji ve Metabolizma

Akademik Unvanlar / Görevler

Prof. Dr., Hacettepe Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2010 - Devam Ediyor

Doç. Dr., Hacettepe Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2005 - 2010

Yrd. Doç. Dr., Hacettepe Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 1998 - 2005

Öğretim Görevlisi Dr., İstanbul Üniversitesi, Sağlık Bilimleri Enstitüsü, Genetik Anabilim Dalı, 1994 - 1998

Uzman, Hacettepe Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 1988 - 1992

Verdiği Dersler

metabolik hastalıklara yaklaşım, Lisans, 2017 - 2018, 2016 - 2017

Kalıtsal metabolik hastalıklar tanı ve izlem, Doktora, 2017 - 2018, 2016 - 2017

malnütrisyon, Lisans, 2017 - 2018

asidosis, Lisans, 2017 - 2018, 2016 - 2017

fenilketonüri, Lisans, 2017 - 2018, 2016 - 2017

Kalıtsal Metabolik hastalıkların moleküler temeli, Doktora, 2017 - 2018, 2016 - 2017

yenidoğan taramalar, Lisans, 2017 - 2018, 2016 - 2017

dislipidemiler, Lisans, 2017 - 2018, 2016 - 2017

SCI, SSCI ve AHCI İndekslerine Giren Dergilerde Yayınlanan Makaleler

- I. **Long-term clinical evaluation of patients with alpha-mannosidosis – A multicenter study**
KÖSE E., KASAPKARA Ç. S., İNCİ A., YILDIZ Y., Sürücü Kara İ., Kahraman A. B., TÜMER L., DURSUN A., EMİNOĞLU F. T.
European Journal of Medical Genetics, cilt.68, 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 : a journal of neurology, cilt.147, sa.1, ss.311-324, 2024 (SCI-Expanded)
- III. **Predictors of eventual requirement of phenylalanine-restricted diet in young infants with phenylalanine hydroxylase deficiency initially managed with sapropterin monotherapy**
ÇIKI K., YILDIZ Y., KAHRAMAN A. B., ÖZGÜL R. K., COŞKUN T., DURSUN A., TOKATLI A., SİVRİ S.
Molecular Genetics and Metabolism, cilt.140, sa.3, 2023 (SCI-Expanded)
- IV. **COVID-19 in inherited metabolic disorders: Clinical features and risk factors for disease severity**
KAHRAMAN A. B., YILDIZ Y., ÇIKI K., Erdal I., AKAR H. T., DURSUN A., TOKATLI A., SİVRİ S.
Molecular Genetics and Metabolism, cilt.139, sa.2, 2023 (SCI-Expanded)
- V. **Organic acidemias in the neonatal period: 30 years of experience in a referral center for inborn errors of metabolism**
ÜNSAL Y., YURDAKÖK M., YİĞİT Ş., ÇELİK H. T., DURSUN A., SİVRİ H. S., TOKATLI A., COŞKUN T.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, cilt.35, sa.11, ss.1345-1356, 2022 (SCI-Expanded)
- VI. **An investigation of different intracellular parameters for Inborn Errors of Metabolism: Cellular stress, antioxidant response and autophagy**
Vardar Acar N., DURSUN A., Aygün D., Gürses Cila H. E., LAY İ., GÜLBAKAN B., ÖZGÜL R. K.
Free Radical Biology and Medicine, cilt.179, ss.190-199, 2022 (SCI-Expanded)
- VII. **Biallelic mutations in ELFN1 gene associated with developmental and epileptic encephalopathy and joint laxity**
DURSUN A., YALNIZOĞLU D., Yılmaz D. Y., Oguz K. K., GÜLBAKAN B., KOŞUKCU C., AKAR H. T., Kahraman A. B., Acar N. V., GÜNBEY C., et al.
EUROPEAN JOURNAL OF MEDICAL GENETICS, cilt.64, sa.11, 2021 (SCI-Expanded)
- VIII. **Clinical and molecular characteristics of carnitine-acylcarnitine translocase deficiency with c.270delC and a novel c.408C>A variant**
BİLGİNER GÜRBÜZ B., YÜCEL YILMAZ D., ÖZGÜL R. K., KOŞUKCU C., DURSUN A., SİVRİ H. S., COŞKUN T., TOKATLI A.
TURKISH JOURNAL OF PEDIATRICS, cilt.63, sa.4, ss.691-696, 2021 (SCI-Expanded)
- IX. **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, cilt.63, sa.11, 2020 (SCI-Expanded)
- X. **Predictors of acute metabolic decompensation in children with maple syrup urine disease at the emergency department**
YILDIZ Y., Akcan Yıldız L., DURSUN A., TOKATLI A., COŞKUN T., TEKŞAM Ö., SİVRİ H. S.
EUROPEAN JOURNAL OF PEDIATRICS, cilt.179, sa.7, ss.1107-1114, 2020 (SCI-Expanded)
- XI. **Genotypes and estimated prevalence of phosphomannomutase 2 deficiency in Turkey differ significantly from those in Europe**
YILDIZ Y., Arslan M., Celik G., Kasapkara C. S., Ceylaner S., DURSUN A., SİVRİ H. S., COŞKUN T., TOKATLI A.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.182, sa.4, ss.705-712, 2020 (SCI-Expanded)
- XII. **Oral health status of children with phenylketonuria**
BALLIKAYA E., YILDIZ Y., SİVRİ H. S., TOKATLI A., DURSUN A., Olmez S., COŞKUN T., Tekcicek M. U.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, cilt.33, sa.3, ss.361-365, 2020 (SCI-Expanded)
- XIII. **Presentation of 14 alkaptonuria patients from Turkey**
AKBABA A. İ., ÖZGÜL R. K., DURSUN A.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, cilt.33, sa.2, ss.289-294, 2020 (SCI-Expanded)
- XIV. **The effectiveness of enzyme replacement therapy on cardiac findings in patients with**

mucopolysaccharidosis

BİLGİNER GÜRBÜZ B., AYPAR E., COŞKUN T., ALEHAN D., DURSUN A., TOKATLI A., SİVRİ H. S.

JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, cilt.32, sa.10, ss.1049-1053, 2019 (SCI-Expanded)

- XV. **Determinants of Riboflavin Responsiveness in Multiple Acyl-CoA Dehydrogenase Deficiency**
YILDIZ Y., TALİM B., Haliloglu G., Topaloglu H., AKÇÖREN Z., DURSUN A., SİVRİ H. S., COŞKUN T., TOKATLI A.
PEDIATRIC NEUROLOGY, cilt.99, ss.69-75, 2019 (SCI-Expanded)
- XVI. **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 (SCI-Expanded)
- XVII. **Imaging liver nodules in tyrosinemia type-1: A retrospective review of 16 cases in a tertiary pediatric hospital**
ÖZCAN H. N., KARÇAALTINCABA M., Pektas E., SİVRİ H. S., OĞUZ B., DURSUN A., TOKATLI A., COŞKUN T., HALİLOĞLU M.
EUROPEAN JOURNAL OF RADIOLOGY, cilt.116, ss.41-46, 2019 (SCI-Expanded)
- XVIII. **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, cilt.42, sa.2, ss.381-388, 2019 (SCI-Expanded)
- XIX. **The genotypic and phenotypic spectrum of MTO1 deficiency**
O'Byrne J. J., Tarailo-Graovac M., Ghani A., Champion M., Deshpande C., DURSUN A., ÖZGÜL R. K., Freisinger P., Garber I., Haack T. B., et al.
MOLECULAR GENETICS AND METABOLISM, cilt.123, sa.1, ss.28-42, 2018 (SCI-Expanded)
- XX. **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, cilt.173, sa.11, ss.2954-2967, 2017 (SCI-Expanded)
- XXI. **Deoxyguanosine kinase deficiency: a report of four patients**
Unal O., ÖZTÜRK HİŞMİ B., KILIÇ M., HIZARCIOĞLU GÜLŞEN H., COŞKUN T., Sivri S. H., DURSUN A., YÜCE A., TOKATLI A.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, cilt.30, sa.6, ss.697-702, 2017 (SCI-Expanded)
- XXII. **Novel and prevalent CYP11B1 gene mutations in Turkish patients with 11-beta hydroxylase deficiency**
KANDEMİR N., Yilmaz D. Y., GÖNÇ E. N., Ozon A., ALİKAŞİFOĞLU A., DURSUN A., ÖZGÜL R. K.
JOURNAL OF STEROID BIOCHEMISTRY AND MOLECULAR BIOLOGY, cilt.165, ss.57-63, 2017 (SCI-Expanded)
- XXIII. **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, cilt.26, sa.1, ss.1-12, 2017 (SCI-Expanded)
- XXIV. **Partial hydatidiform mole in a phenylketonuria patient treated with sapropterin dihydrochloride**
YILDIZ Y., DURSUN A., TOKATLI A., COŞKUN T., Sivri S.
GYNECOLOGICAL ENDOCRINOLOGY, cilt.33, sa.1, ss.19-20, 2017 (SCI-Expanded)
- XXV. **A Nonvirilized form of Classic 3 beta-Hydroxysteroid Dehydrogenase Deficiency Due to a Homozygous S218P Mutation in the HSD3B2 Gene in a Girl with Classic Phenylketonuria**
ALİKAŞİFOĞLU A., Buyukyilmaz G., GÖNÇ E. N., ÖZÖN Z. A., KANDEMİR N., DÜNDAR M., POLAT S., PEKTAŞ E., DURSUN A., Sivri S., et al.
HORMONE RESEARCH IN PAEDIATRICS, cilt.86, ss.281, 2016 (SCI-Expanded)
- XXVI. **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, cilt.46, sa.2, ss.404-408, 2016 (SCI-Expanded)

- XXVII. **Late-diagnosed phenylketonuria in an eight-year-old boy with dyslexia and attention-deficit hyperactivity disorder**
YILDIZ Y., DURSUN A., TOKATLI A., COŞKUN T., SİVRİ H. S.
TURKISH JOURNAL OF PEDIATRICS, cilt.58, sa.1, ss.94-96, 2016 (SCI-Expanded)
- XXVIII. **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, ss.213-218, 2015 (SCI-Expanded)
- XXIX. **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, cilt.174, sa.8, ss.1077-1084, 2015 (SCI-Expanded)
- XXX. **Ailevi Hiperkolesterolemili Hastaların Mutasyon Analiz Sonuçlarının Simone-Broome Kriterleriyle Değerlendirilmesi**
AYKAN H. H., ÖZGÜL R. K., Güzel A., COŞKUN T., DURSUN A.
Türkiye Çocuk Hastalıkları Dergisi, cilt.9, sa.3, ss.176-183, 2015 (SCI-Expanded)
- XXXI. **Biallelic mutations in SNX14 cause a syndromic form of cerebellar atrophy and lysosome-autophagosome dysfunction**
AKIZU N., CANTAGREL V., ZAKI M. S., Al-Gazali L., WANG X., ROSTI R. O., DIKOGLU E., GELOT A. B., ROSTI B., VAUX K. K., et al.
NATURE GENETICS, cilt.47, sa.5, ss.528-536, 2015 (SCI-Expanded)
- XXXII. **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, cilt.57, ss.388-393, 2015 (SCI-Expanded)
- XXXIII. **Phenotypic and genotypic spectrum of Turkish patients with isovaleric acidemia**
ÖZGÜL R. K., Karaca M., Kilic M., KUCUK O., YUCEL-YILMAZ D., ÜNAL O., HISMI B., Aliefendioglu D., SIVRI S., TOKATLI A., et al.
EUROPEAN JOURNAL OF MEDICAL GENETICS, cilt.57, sa.10, ss.596-601, 2014 (SCI-Expanded)
- XXXIV. **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, cilt.34, sa.3, ss.265-266, 2014 (SCI-Expanded)
- XXXV. **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, cilt.534, sa.2, ss.197-203, 2014 (SCI-Expanded)
- XXXVI. **Pregnancy and Lactation Outcomes in a Turkish Patient with Lysinuric Protein Intolerance**
Unal O., COŞKUN T., ORHAN D., Tokatli A., DURSUN A., ÖZTÜRK HİŞMİ B., ÖZYÜNCÜ Ö., Sivri S. H. K.
JIMD REPORTS, VOL 13, cilt.13, ss.33-36, 2014 (SCI-Expanded)
- XXXVII. **A Patient With Pyruvate Carboxylase Deficiency and Nemaline Rods on Muscle Biopsy**
Unal O., ORHAN D., Ostergaard E., TOKATLI A., DURSUN A., ÖZTÜRK HİŞMİ B., COŞKUN T., Wibrand F., Kalkanoglu-Sivri H. S.
JOURNAL OF CHILD NEUROLOGY, cilt.28, sa.11, ss.1505-1508, 2013 (SCI-Expanded)
- XXXVIII. **Vanishing White Matter With Hepatomegaly and Hypertriglyceridemia Attacks**
Unal O., ÖZGEN MOCAN B., ORHAN D., TOKATLI A., Hismi B. O., DURSUN A., COŞKUN T., Kalkanoglu-Sivri H. S.
JOURNAL OF CHILD NEUROLOGY, cilt.28, sa.11, ss.1509-1512, 2013 (SCI-Expanded)
- XXXIX. **Cobalamin C defect: a patient of late-onset type with homozygous p. R132*mutation**
KILIÇ M., ÖZGÜL R. K., DURSUN A., TOKATLI A., Kalkanoglu-Sivri H. S., Anlar B., Fowler B., COŞKUN T.
TURKISH JOURNAL OF PEDIATRICS, cilt.55, sa.6, ss.633-636, 2013 (SCI-Expanded)
- XL. **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, cilt.58, sa.10, ss.675-678, 2013 (SCI-Expanded)

- XLII. **Molecular and clinical evaluation of Turkish patients with lysinuric protein intolerance**
Guzel-Ozanturk A., ÖZGÜL R. K., Unal O., Hismi B., Aydin H. I., Sivri S., TOKATLI A., COŞKUN T., Aksoz E., DURSUN A.
GENE, cilt.521, sa.2, ss.293-295, 2013 (SCI-Expanded)
- XLIII. **Novel Mutations in the PC Gene in Patients with Type B Pyruvate Carboxylase Deficiency**
Ostergaard E., Duno M., Moller L. B., Kalkanoglu-Sivri H. S., DURSUN A., ALİEFENDİOĞLU D., Leth H., Dahl M., Christensen E., Wibrand F.
JIMD REPORTS - CASE AND RESEARCH REPORTS, 2012/6, cilt.9, ss.1-5, 2013 (SCI-Expanded)
- XLIV. **ACUTE INTERMITTENT PORPHYRIA: STILL UNCALLED BY PHYSICIANS**
ÖZTÜRK HİŞMİ B., Tanriover M. D., Unal O., Sener E., TEMUÇİN Ç. M., Sivri H. S., DURSUN A., Coskun T.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.35, 2012 (SCI-Expanded)
- XLV. **MOLECULAR CHARACTERISATION OF BIOTINIDASE GENE MUTATIONS IN TURKISH PATIENTS; AN UPDATE OF THE RESULTS**
Karaca M., Yucel D., Unal O., Guzel A., TOKATLI A., COŞKUN T., DURSUN A., SİVRİ H. S., ÖZGÜL R. K.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.35, 2012 (SCI-Expanded)
- XLVI. **ENZYME REPLACEMENT THERAPY (ERT) RESULTS OF PATIENTS WITH MUCOPOLYSACCHARIDOSIS TYPE II (HUNTER SYNDROME)**
Unal O., Dogru-Ersoz D., Alehan D., Saglam M., Hismi B., DURSUN A., Tokatli A., Coskun T., Sivri H. S.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.35, 2012 (SCI-Expanded)
- XLVII. **A PATIENT WITH PYRUVATE CARBOXYLASE DEFICIENCY AND NEMALINE RODS ON MUSCLE BIOPSY**
Unal O., ÖZTÜRK HİŞMİ B., DURSUN A., Tokatli A., Coskun T., Wibrand F., Sivri H. S.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.35, 2012 (SCI-Expanded)
- XLVIII. **DIETARY MANAGEMENT AND GROWTH OF TETRAHYDROBIOPTERIN RESPONSIVE TURKISH PKU PATIENTS**
Gokmen-Ozel H., Unal O., Kalkanoglu-Sivri H. S., Koksall G., DURSUN A., Tokatli A., Coskun T.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.35, 2012 (SCI-Expanded)
- XLIX. **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, cilt.35, 2012 (SCI-Expanded)
- L. **FEVER OF UNKNOWN ORIGIN IN A YOUNG ADULT WITH END-STAGE RENAL DISEASE, PREMATURE CORONARY ARTERY DISEASE AND POLYNEUROPATHY**
ÖZTÜRK HİŞMİ B., Yasar Y., Unal O., Kilic L., Turkmen E., Erdem Y., DURSUN A., Sivri H. S., Coskun T.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.35, 2012 (SCI-Expanded)
- LI. **PROPIONIC ACIDEMIA PRESENTING WITH PERSISTENT PULMONARY HYPERTENSION IN TWO NEONATES**
Hisim B., TEKŞAM Ö., Unal O., Takci S., Ertugrul İ., SİVRİ H. S., DURSUN A., TOKATLI A., COŞKUN T.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.35, 2012 (SCI-Expanded)
- LII. **Methylmalonic acidemia mimicking diabetic ketoacidosis in an infant**
Güven A., Cebeci N., DURSUN A., Aktekin E. H., Baumgartner M. R., Fowler B.
PEDIATRIC DIABETES, cilt.13, sa.6, 2012 (SCI-Expanded)
- LIII. **MOLYBDENUM COFACTOR (MOCO) DEFICIENCY TYPE B; CLINICAL, BIOCHEMICAL AND NEUROIMAGING FEATURES OF FIVE PATIENTS WITH TWO NOVEL MUTATIONS**
ÖZTÜRK HİŞMİ B., Unal O., Sass J. O., Beermann F., Ichida K., DURSUN A., Sivri H. S., Tokatli A., Coskun T.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.35, 2012 (SCI-Expanded)
- LIV. **SEVERE AZOTEMIA AND HYPERNATREMIC DEHYDRATION IN AN INFANT WITH PHENYLKETONURIA**
Unal O., DÜZOVA A., ÖZTÜRK HİŞMİ B., DURSUN A., Tokatli A., Coskun T., Sivri H. S.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.35, 2012 (SCI-Expanded)
- LIV. **Detection of other inborn errors of metabolism in hyperphenylalaninemic babies picked up on narrow-spectrum screening programs**

Unal O., ÖZTÜRK HİŞMİ B., COŞKUN T., TOKATLI A., DURSUN A., SİVRİ H. S.

TURKISH JOURNAL OF PEDIATRICS, cilt.54, sa.4, ss.409-412, 2012 (SCI-Expanded)

- LV. Identification of a Novel Twinkle Mutation in a Family With Infantile Onset Spinocerebellar Ataxia by Whole Exome Sequencing**
DUNDAR H., ÖZGÜL R. K., YALNIZOĞLU D., Erdem S., Oguz K. K., Tuncel D., TEMUÇİN Ç. M., DURSUN A.
PEDIATRIC NEUROLOGY, cilt.46, sa.3, ss.172-177, 2012 (SCI-Expanded)
- LVI. A Zinc Sulphate-Resistant Acrodermatitis Enteropathica Patient with a Novel Mutation in SLC39A4 Gene**
Kilic M., Taskesen M., Coskun T., Gurakan F., Tokatli A., Sivri H. S., DURSUN A., Schmitt S., Kury S.
JIMD REPORTS - CASE AND RESEARCH REPORTS, 2011/2, cilt.2, ss.25-28, 2012 (SCI-Expanded)
- LVII. Identification of Mutations and Evaluation of Cardiomyopathy in Turkish Patients with Primary Carnitine Deficiency**
KILIÇ M., ÖZGÜL R. K., COŞKUN T., Yucel D., KARACA M. A., SİVRİ H. S., TOKATLI A., ŞAHİN M., KARAGÖZ T., DURSUN A.
JIMD REPORTS - CASE AND RESEARCH REPORTS, 2011/3, cilt.3, ss.17-23, 2012 (SCI-Expanded)
- LVIII. IDENTIFICATION OF NOVEL MUTATIONS IN PROMOTER AND CODING REGIONS IN ALDOB GENE CAUSING HEREDITARY FRUCTOSE INTOLERANCE**
Yucel D., ÖZGÜL R. K., COŞKUN T., SİVRİ H. S., TOKATLI A., DURSUN A.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.34, 2011 (SCI-Expanded)
- LIX. HEMOPHAGOCYTOSIS IN THREE PATIENTS WITH ORGANIC ACIDEMIA INVOLVING PROPIONATE METABOLISM**
Gokce M., Unal O., ÖZTÜRK HİŞMİ B., Gumruk F., Coskun T., DURSUN A., Tokatli A., Sivri K. H. S.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.34, 2011 (SCI-Expanded)
- LX. OUTCOME OF ENZYME REPLACEMENT THERAPY IN PATIENTS WITH MUCOPOLYSACCHARIDOSIS TYPE VI**
Kilic M., Kalkanoglu-Sivri H. S., DURSUN A., Tokatli A., Coskun T.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.34, 2011 (SCI-Expanded)
- LXI. A novel mutation in the DGUOK gene in a Turkish newborn with mitochondrial depletion syndrome**
KILIÇ M., SİVRİ H. S., DURSUN A., TOKATLI A., De Meirleir L., Seneca S., AKÇÖREN Z., YİĞİT Ş., Topaloglu H., COŞKUN T.
TURKISH JOURNAL OF PEDIATRICS, cilt.53, sa.1, ss.79-82, 2011 (SCI-Expanded)
- LXII. THE CARDIAC MANIFESTATION AND RESPONSE TO L-CARNITINE TREATMENT IN 14 CASES WITH PRIMARY SYSTEMIC CARNITINE DEFICIENCY: CORRELATION WITH GENOTYPE**
Balci M. C., Yucel D., Ergul Y., ÖZGÜL R. K., Baykal T., Aktuglu-Zeybek C., Ersoy M., Demirkol M., Eker-Omeroglu R., DURSUN A., et al.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.34, 2011 (SCI-Expanded)
- LXIII. CHALLENGES IN THE TREATMENT OF A PATIENT AFFECTED BY BOTH ARGININOSUCCINIC ACIDURIA AND METHYLMALONIC ACIDURIA**
Unal O., ÖZTÜRK HİŞMİ B., Coskun T., Tokatli A., DURSUN A., Sivri K. H. S.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.34, 2011 (SCI-Expanded)
- LXIV. A ZINC SULPHATE-RESISTANT ACRODERMATITIS ENTEROPATHICA PATIENT WITH A NOVEL MUTATION IN SLC39A4 GENE**
Kilic M., Taskesen M., Coskun T., Gurakan F., Tokatli A., Kalkanoglu-Sivri H. S., DURSUN A., Schmitt S., Kury S.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.34, 2011 (SCI-Expanded)
- LXV. THE PRESENCE OF NEGATIVE CORRELATION BETWEEN BLOOD HEMOGLOBIN AND PROPIONYL-CARNITINE LEVELS IN METHYLMALONIC AND PROPIONIC ACIDEMIAS**
Aydin H., Sivri S., DURSUN A., Tokatli A., Kutuk G., Onol S., Coskun T.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.34, 2011 (SCI-Expanded)
- LXVI. MUTATION ANALYSIS IN ARSB GENE IN TURKISH PATIENTS WITH MPS TYPE VI: HIGH PREVALENCE OF L321P MUTATION**
ÖZGÜL R. K., Karaca M., SİVRİ H. S., TOKATLI A., COŞKUN T., DURSUN A.

- JOURNAL OF INHERITED METABOLIC DISEASE, cilt.34, 2011 (SCI-Expanded)
- LXVII. **PHENOTYPICAL PROPERTIES AND RESPONSE TO CHOLESTEROL THERAPY OF SMITH-LEMLI-OPITZ SYNDROME CASES**
Kilic M., Tokatli A., ALANAY Y., Kilic E., Kalkanoglu-Sivri H. S., DURSUN A., Onol S., Haliloglu G., Utine G. E., BODUROĞLU O. K., et al.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.34, 2011 (SCI-Expanded)
- LXVIII. **Mutation Spectrum of Fumarylacetoacetase Gene and Clinical Aspects of Tyrosinemia Type I Disease**
DURSUN A., ÖZGÜL R. K., Sivri S., TOKATLI A., Guzel A., Mesci L., KILIÇ M., Aliefendioglu D., Ozcay F., Gunduz M., et al.
JIMD REPORTS: CASE AND RESEARCH REPORTS, 2011/1, cilt.1, ss.17-21, 2011 (SCI-Expanded)
- LXIX. **Mutations in the G6PC3 Gene Cause Dursun Syndrome**
Banka S., Newman W. G., ÖZGÜL R. K., DURSUN A.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, sa.10, ss.2609-2611, 2010 (SCI-Expanded)
- LXX. **HACETTEPE EXPERIENCE WITH PEROXISOMAL DISORDERS UNDER FOUR YEARS OF AGE**
Kilic M., Tokatli A., Sivri H. S., DURSUN A., Topaloglu H., Wanders R. J. A., Coskun T.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)
- LXXI. **MUTATION PROFILE OF BCKDHA, BCKDHB AND DBT GENES FOR MAPLE SYRUP URINE DISEASE IN TURKEY**
Ozgul R. K., Guzel A., Dundar H., Yucel D., Yilmaz A., Unal O., Tokatli A., Sivri H. S., Coskun T., DURSUN A.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)
- LXXII. **A TURKISH PATIENT WITH LATE ONSET cbIC DEFECT CAUSED BY c.394C > T MUTATION**
Kilic M., DURSUN A., Tokatli A., Sivri H. S., Anlar B., Fowler B., Coskun T.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)
- LXXIII. **SCREENING OF ATP7B GENE MUTATIONS IN TURKISH PATIENTS WITH WILSON DISEASE BY CUSTOM DESIGNED RESEQUENCING MICROARRAYS**
Yilmaz A., Guzel A., Dundar H., DURSUN A., Uslu N., Yuce A., Ozgul R. K.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)
- LXXIV. **NOVEL HOMOGENITISATE DIOXIGENASE (HGD) GENE MUTATIONS IN ALKAPTONURIA PATIENTS**
Hatipoglu E., Ozgul R. K., Sivri H. S., Coskun T., Tokatli A., Karaca S., Kucuk O., Kilic M., Akcelik M., DURSUN A.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)
- LXXV. **SIX NOVEL MUTATIONS IN TURKISH PATIENTS WITH ISOVALERIC ACIDEMIA**
Kucuk O., Ozgul R. K., Karaca M., Sivri H. S., Coskun T., Tokatli A., Hatipoglu E., Unal O., Akcelik M., DURSUN A.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)
- LXXVI. **MUTATIONS IN FUMARYLACETOACETATE HYDROLASE GENE AND GENOTYPE-PHENOTYPE RELATION**
Ozgul R. K., Guzel A., Mesci L., Sivri H. S., Kilic M., Ozcay F., Gunduz M., Aydin H., Aliefendioglu D., Coskun T., et al.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)
- LXXVII. **EVALUTION OF 42 PATIENTS WITH HYPERPHENYLALANINEMIA CAUSED BY A DEFECT IN TETRAHYDROBIOPTERIN METABOLISM**
Kizilelma A., Tokatli A., Kalkanoglu-Sivri H. S., DURSUN A., Aydin H., Blau N., Coskun T.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)
- LXXVIII. **NOVEL MUTATIONS IN TURKISH PATIENTS WITH PRIMARY CARNITINE DEFICIENCY**
Kilic M., Ozgul R. K., Yucel D., Karaca M., DURSUN A., Sivri H. S., Tokatli A., Sahin M., Karagoz T., Coskun T.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)
- LXXIX. **THE SPECTRUM AND FREQUENCY OF ALDOLASE B GENE MUTATIONS IN TURKISH PATIENTS WITH HEREDITARY FRUCTOSE INTOLERANCE**
Yucel D., Ozgul R. K., Yilmaz A., Sivri H. S., Coskun T., Unal O., Tokatli A., DURSUN A.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)
- LXXX. **INFANTILE REFSUM DISEASE IN A TURKISH PATIENT: CASE REPORT**
Kilic M., Karli O. K., Coskun T., Haliloglu G., Wanders R. J. A., DURSUN A., Sivri H. S., Tokatli A., Topcu M.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)
- LXXXI. **GALACTOSEMIA IN A TURKISH POPULATION WITH A HIGH PREVALENCE OF Q188R MUTATION**
Guzel A., Ozgul R. K., Dundar H., Coskun T., Sivri H. S., Tokatli A., Goksun E., ÖZTÜRK HİŞMİ B., DURSUN A.

- JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)
- LXXXII. **GROWTH AND PROTEIN INTAKE IN PHENYLKETONURIA: RESULTS OF 398 TURKISH CHILDREN**
GOKMEN-OZEL H., Buyuktuncer Z., KOKSAL G., KILIÇ M., DURSUN A., KALKANOGLU-SIVRI S., TOKATLI A., COŞKUN T.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)
- LXXXIII. **TRANSCOBALAMIN II DEFICIENCY IN TWO CASES WITH A NOVEL MUTATION**
ÜNAL S., Ozgul R. K., DURSUN A., Yetgin S., Coskun T., Rupar T., Cetin M.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)
- LXXXIV. **A NOVEL MUTATION IN BETA KETOTHIOLASE DEFICIENCY**
Unal O., ÖZTÜRK HİŞMİ B., Kilic M., DURSUN A., Kalkanoglu-Sivri H. S., Tokatli A., Coskun T., Sass O.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)
- LXXXV. **CIRRHOSIS ASSOCIATED WITH PROPIONATE METABOLISM**
DURSUN A., Dundar H., Ozgul R. K., Talim B., Kale G., Demir H., Temizel S., Tokatli A., Sivri H. S., Coskun T.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)
- LXXXVI. **MOLECULAR AND STRUCTURAL ANALYSIS OF SIX NONSENSE MUTATIONS IN MUT METHYLMALONIC ACIDEMIA PATIENTS INCLUDING TWO NOVEL NONSENSE MUTATIONS**
Dundar H., Ozgul R. K., Unal O., Karaca M., Aydin H., Tokatli A., Sivri H. S., Coskun T., DURSUN A.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)
- LXXXVII. **THREE CASES WITH FRUCTOSE 1,6-BISPHOSPHATASE DEFICIENCY: TWO NOVEL MUTATIONS**
Yucel D., Ozgul R. K., Tokatli A., Sivri H. S., Guzel A., Coskun T., DURSUN A.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)
- LXXXVIII. **POLYNEUROPATHY AS THE MAIN PRESENTING SYMPTOM IN PDH DEFICIENCY**
Unal O., ÖZTÜRK HİŞMİ B., Kilie M., DURSUN A., Kalkanoglu-Sivri H. S., Tokatli A., Coskun T., Zeviani M.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)
- LXXXIX. **A NOVEL MUTATION IN DGUOK GENE IN A TURKISH NEWBORN**
Kilic M., DURSUN A., Sivri H. S., Tokatli A., Akcoren Z., Yigit S., Vezir E., Seneca S., Demerlier L., Coskun T.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)
- XC. **MUTATIONS IN UBIQUITOUSLY EXPRESSED GLUCOSE-6-PHOSPHATASE CATALYTIC SUBUNIT (G6PC3) CAUSE DURSUN SYNDROME**
Banka S., Newman W. G., Donnai D., Crow Y. J., Chervinsky E., Shalev S., Yeganeh S., Ozgul R. K., DURSUN A.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)
- XCII. **ASSOCIATION OF POLYNEUROPATHY, MENTAL RETARDATION, SENSORINEURAL HEARING LOSS, 6th NERVE PALSY, CONVULSIONS, AND ORAL DYSKINESIA; A PROPABLE NEW NEUROMETABOLIC DISORDER**
DURSUN A., YALNIZOĞLU D., DÜNDAR H., ERDEM S., AKARSU A. N., ÖZGÜL R. K.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)
- XCIII. **IDENTIFICATION OF MUTATIONS IN THE PCCA AND PCCB GENES CAUSING PROPIONIC ACIDEMIA IN TURKISH PATIENTS**
Ozgul R. K., Yucel D., ÖZTÜRK HİŞMİ B., Karaca M., Sivri H. S., Coskun T., Tokatli A., DURSUN A.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)
- XCIII. **RENAL AGENESIS IN ASSOCIATION WITH MATERNAL PKU SYNDROME**
Kilic M., Sivri H. S., Tokatli A., DURSUN A., Coskun T.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)
- XCIV. **MUTATION DETECTION IN TURKISH PATIENTS WITH GLUTARIC ACIDURIA TYPE I**
Guzel A., Ozgul R. K., Yucel D., Karaca M., Kilic M., Coskun T., Tokatli A., Sivri H. S., DURSUN A.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)
- XCV. **GENOME-WIDE GENOTYPING FOR THE CHARACTERIZATION OF DISEASE LOCUS IN A FAMILY WITH AN UNCHARACTERIZED NEUROMETABOLIC DISEASE**
Dundar H., Yucel D., DURSUN A., Ozgul R. K.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)
- XCVI. **VITAMIN B6 AND B12 STATUS IN TURKISH CHILDREN WITH PHENYLKETONURIA**

Buyuktuncer Z., GOKMEN-OZEL H., KUCUKKASAP T., KOKSAL G., KILIÇ M., DURSUN A., KALKANOĞLU-SIVRI H. S., TOKATLI A., COŞKUN T., BESLER H. T.

JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)

- XCVII. Von Willebrand disease associated with heterozygous factor V G1691A mutation and thrombosis in a patient with mut(0) methylmalonic acidemia: a paradoxical phenomenon**
Aytac S., ÜNAL S., Coskun T., Tokatli A., Sivri S., DURSUN A., Cila A., Gurgey A., Gumruk F.
HAEMOPHILIA, cilt.16, sa.2, ss.408, 2010 (SCI-Expanded)
- XCVIII. Four novel PDHA1 mutations in pyruvate dehydrogenase deficiency**
Ostergaard E., Moller L. B., Kalkanoglu-Sivri H. S., DURSUN A., Kibaek M., Thelle T., Christensen E., Duno M., Wibrand F.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.32, sa.1, 2009 (SCI-Expanded)
- XCIX. Zellweger syndrome with unusual findings: non-immune hydrops fetalis, dermal erythropoiesis and hypoplastic toe nails**
DURSUN A., Gucer S., Ebberink M. S., YİĞİT Ş., Wanders R. J. A., Waterham H. R.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.32, sa.1, 2009 (SCI-Expanded)
- C. 3-phosphoglycerate dehydrogenase deficiency: a case report of a treatable cause of seizures**
COŞKUN T., Aydin H. I., KILIÇ M., DURSUN A., Haliloglu G., Topaloglu H., Karli-Oguz K., de Koning T. J.
TURKISH JOURNAL OF PEDIATRICS, cilt.51, sa.6, ss.587-592, 2009 (SCI-Expanded)
- CI. Increased Frequency of Extremely Skewed X Chromosome Inactivation in Juvenile Idiopathic Arthritis**
Uz E., Mustafa C., TOPALOĞLU R., BİLGİNER Y., DURSUN A., KASAPÇOPUR Ö., ÖZEN S., BAKKALOĞLU A., ÖZÇELİK T.
ARTHRITIS AND RHEUMATISM, cilt.60, sa.11, ss.3410-3412, 2009 (SCI-Expanded)
- CII. Molecular genetics of maple syrup urine disease in the Turkish population**
Gorzelay K., DURSUN A., COŞKUN T., Kalkanoglu-Sivri S. H., GÖKÇAY G. F., Demirkol M., Feyen O., Wendel U.
TURKISH JOURNAL OF PEDIATRICS, cilt.51, sa.2, ss.97-102, 2009 (SCI-Expanded)
- CIII. Familial pulmonary arterial hypertension, leucopenia, and atrial septal defect: a probable new familial syndrome with multisystem involvement**
DURSUN A., ÖZGÜL R. K., SOYDAŞ A., Tugrul T., GÜRGEY A., ÇELİKER A., Barst R. J., Knowles J. A., Mahesh M., Morse J. H.
CLINICAL DYSMORPHOLOGY, cilt.18, sa.1, ss.19-23, 2009 (SCI-Expanded)
- CIV. Renal functions of twenty seven children with methylmalonic Acidemia (MMA): Is there a good marker?**
Aydin H. I., Duzova A., DURSUN A., Sivri S., Aksoy T., Kiratli P., Tokatli A., Bakkaloglu A., Coskun T.
PEDIATRIC NEPHROLOGY, cilt.23, sa.9, ss.1587, 2008 (SCI-Expanded)
- CV. Severe axonal polyneuropathy due to dichloroacetate in a patient with pyruvate dehydrogenase deficiency**
DURSUN A., Tokatly A., Sivri H. S., Coskun T.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.31, ss.63, 2008 (SCI-Expanded)
- CVI. Favourable outcome with oral creatine supplementation: Clinical and laboratory findings in five patients with GAMT deficiency**
Haliloglu G., Karli O. K., DURSUN A., Tokatli A., Bodamer O., Coskun T., Topcu M.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.31, ss.69, 2008 (SCI-Expanded)
- CVII. Pancreatic beta cell reserve and insulin sensitivity in maple syrup urine disease**
DURSUN A., Sarikabadayi Y. U., Ozon Z. A.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.31, ss.7, 2008 (SCI-Expanded)
- CVIII. Argininosuccinic aciduria associated with pancreatitis**
DURSUN A., Sivri H. S., Oezon A., Akcaoren Z., Tokatly A., Koch H. G., Coskun T.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.31, ss.90, 2008 (SCI-Expanded)
- CIX. Renal function in twenty seven children with methylmalonic acidemia (MMA): Is there a good marker?**
Aydin H., DÜZOVA A., Kalkanoglu S. H. S., DURSUN A., Aksoy T., Kiratli P., Tokatli A., Bakkaloglu A., Coskun T., Fowler

B.

JOURNAL OF INHERITED METABOLIC DISEASE, cilt.31, ss.25, 2008 (SCI-Expanded)

- CX. **A Zellweger syndrome associated with non-immune hydrops fetalis, lung hypoplasia and dermal erythropoiesis**
DURSUN A., Doertttepe Y., Gueer S., Yigit S., Wanders R. J. A.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.31, ss.68, 2008 (SCI-Expanded)
- CXI. **Molecular genetics of maple syrup urine disease (MSUD) in Turkish patients**
Gorzelay K., DURSUN A., Coskun T., GÖKÇAY G. F., Demirkol M., Wendel U.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.30, ss.25, 2007 (SCI-Expanded)
- CXII. **Hearing loss in biotinidase deficiency: Genotype-phenotype correlation**
Sivri H. S. K. L., Genc G. A., Tokatly A., DURSUN A., Copkun T., Aydyn H. Y., Lu L. S., Belgin E., Jensen K., Wolf B.
JOURNAL OF PEDIATRICS, cilt.150, sa.4, ss.439-442, 2007 (SCI-Expanded)
- CXIII. **Haematological findings in children with inborn errors of metabolism**
Tavil B., Sivri H. S. K., Coskun T., Gurgey A., Ozyurek E., DURSUN A., Tokatli A., Altay C., Gumruk F.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.29, sa.5, ss.607-611, 2006 (SCI-Expanded)
- CXIV. **Hearing loss in biotinidase deficiency: Preliminary results indicate genotype-phenotype correlation**
Sivri Kalkanoglu H. S., Genc G. A., Sennaroglu L., Aydin H., DURSUN A., Tokatli A., Wolf B., Belgin E., Coskun T.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.29, ss.105, 2006 (SCI-Expanded)
- CXV. **Psychiatric symptoms of mothers of children with phenylketonuria**
Dursun Soncag A., Sivri Kalkanoglu H. S., Cuhadaroglu F., DURSUN A., Aydin H., Tokatli A., Coskun T.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.29, ss.97, 2006 (SCI-Expanded)
- CXVI. **Basal ganglia involvement in childhood: Creatine deficiency syndrome caused by guanidinoacetate methyltransferase (GAMT) deficiency**
Haliloglu G., DURSUN A., Oguz K. K., Yalnyzoglu D., Aydin H. Y., Aysun S., Sivri H. S., Coskun T., Tokatli A.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.29, ss.145, 2006 (SCI-Expanded)
- CXVII. **CARNITINE TOXICITY IN A PATIENT WITH LCHAD DEFICIENCY**
Kalkanoglu H. S., DURSUN A., Aydin H. I., Tokatli A., Spiekerkotter U., Wendel U., Kazik M., Kandemir N., Talim B., Kale G., et al.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.28, ss.104, 2005 (SCI-Expanded)
- CXVIII. **PYRUVATE DEHYDROGENASE DEFICIENCY DUE TO NOVEL AND KNOWN MUTATIONS IN THE E1 ALPHA SUBUNIT**
Wibrand F., Kalkanoglu H. S., DURSUN A., Tokatli A., Coskun T., Lund A. M., Christensen E., Kibaek M., Ostergaard E., Moller L. B.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.28, ss.122, 2005 (SCI-Expanded)
- CXIX. **A boy with spastic paraparesis and Dyspnea**
Kalkanoglu H., DURSUN A., Tokatli A., Coskun T., Karasimav D., Topaloglu H.
JOURNAL OF CHILD NEUROLOGY, cilt.19, sa.5, ss.397-398, 2004 (SCI-Expanded)
- CXX. **Transient nonketotic hyperglycinemia: Two case reports and literature review**
Aliefendioglu D., Aslan A., Coskun T., DURSUN A., Cakmak F., Kesimer M.
PEDIATRIC NEUROLOGY, cilt.28, sa.2, ss.151-155, 2003 (SCI-Expanded)
- CXXI. **Maple syrup urine disease: Mutation analysis in Turkish patients**
DURSUN A., Henneke M., Ozgul K., Gartner J., Coskun T., Tokatli A., Kalkanoglu H., Demirkol M., Wendel U., Ozalp I.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.25, sa.2, ss.89-97, 2002 (SCI-Expanded)
- CXXII. **Mutation analysis in Turkish patients with hereditary fructose intolerance**
DURSUN A., Kalkanoglu H., Coşkun T., TOKATLI A., Bittner R., KOÇAK N., YÜCE A., Ozalp I., Boehme H.
Journal of Inherited Metabolic Disease, cilt.24, sa.5, ss.523-526, 2001 (SCI-Expanded)

Diğer Dergilerde Yayınlanan Makaleler

- I. **Evaluation of Cardiac Findings in Mucopolysaccharidosis Type III Patients**

BİLGİNER GÜRBÜZ B., AYPAR E., ALEHAN D., TOKATLI A., COŞKUN T., DURSUN A., SİVRİ H. S.

JOURNAL OF PEDIATRIC RESEARCH, cilt.8, sa.2, ss.195-201, 2021 (ESCI)

II. **Cardiomyopathy in patients with type 1 tyrosinemia, and the effect of nitisinone treatment on cardiomyopathy**

BİLGİNER GÜRBÜZ B., AYKAN H. H., ÇIKI K., KARAGÖZ T., Sivri S., DURSUN A., TOKATLI A., COŞKUN T.

CUKUROVA MEDICAL JOURNAL, cilt.46, sa.4, ss.1419-1425, 2021 (ESCI)

III. **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, cilt.2, sa.3, 2016 (ESCI)

IV. **A t(5;16) translocation is the likely driver of a syndrome with ambiguous genitalia, facial dysmorphism, intellectual disability, and speech delay**

OZANTÜRK A., Davis E. E., Sabo A., Weiss M. M., Muzny D., Dugan-Perez S., Sitermans E. A., Gibbs R. A., Ozgul K. R., YALNIZOĞLU D., et al.

COLD SPRING HARBOR MOLECULAR CASE STUDIES, cilt.2, sa.2, 2016 (ESCI)

V. **veziküler trafik bozuklukları**

DURSUN A.

türkiye klinikleri, 2016 (Hakemli Dergi)

VI. **Breast Massage and Mastitis in Early Infancy: Case Report**

Alan S., Iskender D., Tezer H., Devrim I., DURSUN A., KARA A.

JOURNAL OF PEDIATRIC INFECTION, cilt.1, sa.3, ss.121-123, 2007 (ESCI)

Kitap & Kitap Bölümleri

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.

JIMD Reports Case and Research Reports 2011 3, , Editör, SPRINGER, 2011

Hakemli Kongre / Sempozyum Bildiri Kitaplarında Yer Alan Yayınlar

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, Hollanda, 3 - 06 Eylül 2019, cilt.42, ss.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, Hollanda, 3 - 06 Eylül 2019, cilt.42, ss.324

III. **Oxysterol levels as oxidative stress biomarkers in organic acidemia patients**

Eraslan Y., Lay İ., Samadi A., Gürbüz B., DURSUN A., SİVRİ H. S., COŞKUN T.

SSIEM 2018: Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, 04 Eylül 2018 - 07 Eylül 2017, cilt.41, ss.37-219

IV. **Mucopolysaccharidosis in a patient with congenital glaucoma**

DURSUN A., gurbuz b. b., TATAR O., SİVRİ H. S., COŞKUN T.

13. International Congress of Inborn Errors of metabolism, 5 - 08 Eylül 2017

- V. **Three-year experience of pediatric physicians with adult inpatient consultations**
YILDIZ Y., PEKTAŞ E., BİLGİNER GÜRBÜZ B., DURSUN A., TOKATLI A., COŞKUN T., SİVRİ H. S.
13th International Congress of Inborn Errors of Metabolism (ICIEM), Rio de Janeiro, Brezilya, 5 Eylül - 08 Mayıs 2017
- VI. **Oxysterol levels in Organic Acidemia patients: Preliminary results.**
ERASLAN Y., LAY İ., SAMADİ A., DURSUN A., SİVRİ H. S., COŞKUN T.
13th International Congress of Inborn Errors of Metabolism, Rio-De-Janeiro, Brezilya, 5 - 08 Eylül 2017, cilt.5
- VII. **Acute Metabolic Decompensations of Branched-Chain Organic Acidemias in the Pediatric Emergency Department: Clinical Presentation and Outcomes**
SİVRİ H. S., YILDIZ Y., AKCAN L., PEKTAŞ E., BİLGİNER GÜRBÜZ B., DURSUN A., TOKATLI A., COŞKUN T., TEKŞAM Ö.
13th International Congress of Inborn Errors of Metabolism, 5 - 08 Eylül 2017
- VIII. **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 Eylül 2017, cilt.5
- IX. **Etilmelonik ensefalopati: vaka sunumu**
PEKTAŞ E., Yoldaş T. Ç., BİLGİNER GÜRBÜZ B., YILDIZ Y., DURSUN A., SİVRİ H. S., TOKATLI A., COŞKUN T.
XIV. Ulusal Metabolik Hastalıklar ve Beslenme Kongresi, Türkiye, 26 - 30 Nisan 2017
- X. **Erişkinlerde kalıtsal metabolik hastalıklar: yatan hasta konsültasyonları ile üç yıllık deneyim**
YILDIZ Y., PEKTAŞ E., BİLGİNER GÜRBÜZ B., DURSUN A., TOKATLI A., COŞKUN T., SİVRİ H. S.
XIV. Ulusal Metabolik Hastalıklar ve Beslenme Kongresi, Türkiye, 26 - 30 Nisan 2017
- XI. **Fenilketonürlü bireylerin diyetle fenilalanin ve protein alımları: Önerilere uyum nasıldır?**
YILMAZ Ö., BİLGİNER GÜRBÜZ B., YILDIZ Y., GÖKSOY E., DURSUN A., SİVRİ H. S., TOKATLI A., COŞKUN T., GÖKMEN ÖZEL H.
XIV Ulusal Metabolik Hastalıklar ve Beslenme Kongresi, Muğla, Türkiye, 26 - 30 Nisan 2017, ss.152
- XII. **Fenilketonürlü bireylerde Türkiye'ye Özgü Beslenme Rehberi'ne göre enerji ve bazı besin öğeleri alımının değerlendirilmesi**
YILMAZ Ö., BİLGİNER GÜRBÜZ B., YILDIZ Y., GÖKSOY E., DURSUN A., SİVRİ H. S., TOKATLI A., COŞKUN T., GÖKMEN ÖZEL H.
XIV Ulusal Metabolik Hastalıklar ve Beslenme Kongresi, Muğla, Türkiye, 26 - 30 Nisan 2017, ss.151
- XIII. **Fenilketonürlü bireylerde diyet enerji ve protein alımlarının antropometrik ölçümlere etkisi var mıdır?**
YILMAZ Ö., BİLGİNER GÜRBÜZ B., YILDIZ Y., GÖKSOY E., DURSUN A., SİVRİ H. S., TOKATLI A., COŞKUN T., GÖKMEN ÖZEL H.
XIV Ulusal Metabolik Hastalıklar ve Beslenme Kongresi, Muğla, Türkiye, 26 - 30 Nisan 2017, ss.149
- XIV. **Fenilketonürlü bireylerde beslenme ve diyet hasta destek programının değerlendirilmesi**
GÖKMEN ÖZEL H., YILMAZ Ö., YILDIZ Y., GÖKSOY E., BİLGİNER GÜRBÜZ B., DURSUN A., SİVRİ H. S., TOKATLI A., COŞKUN T.
XIV Ulusal Metabolik Hastalıklar ve Beslenme Kongresi, Muğla, Türkiye, 26 - 30 Nisan 2017, ss.148
- XV. **Fenilketonürlü bireylerde besin gruplarının enerji, protein ve fenilalanin alımına katkısı**
YILMAZ Ö., BİLGİNER GÜRBÜZ B., YILDIZ Y., GÖKSOY E., DURSUN A., SİVRİ H. S., TOKATLI A., COŞKUN T., GÖKMEN ÖZEL H.
XIV Ulusal Metabolik Hastalıklar ve Beslenme Kongresi, Muğla, Türkiye, 26 - 30 Nisan 2017, ss.150
- XVI. **adult form metachromatic leucodistrohy caused by a novel mutation**
BİLGİNER GÜRBÜZ B., khasiyer f., KARLI OĞUZ H. K., karabudak r., DURSUN A., TOKATLI A., COŞKUN T., SİVRİ H. S.
MEMG 13, 26 - 30 Ekim 2016
- XVII. **a rare form of mucopolysaccharidosis**
BİLGİNER GÜRBÜZ B., PEKTAŞ E., DURSUN A., COŞKUN T., TOKATLI A.
MEMG 13, 26 - 30 Ekim 2016
- XVIII. **A27 Prenatal findings and autopsy examination in a newborn with multiple acyl CoA dehydrogenase deficiency Abstract Book A27 p 85**

TOKATLI A., YILDIZ Y., DOKUZBOY SIRMA R., YIĞİT Ş., TALİM B., CEYLANER S., PEKTAŞ E., BİLGİNER GÜRBÜZ B., DURSUN A., SİVRİ H. S., et al.

13th Middle East Metabolic Group Meeting, 6, Amman-Jordan, 28 - 30 Ekim 2016, ss.85

- XIX. **presentation of classical galaktosemia with positive newborn screening**
DURSUN A., bozat a., BİLGİNER GÜRBÜZ B., PEKTAŞ E., SİVRİ H. S., TOKATLI A., COŞKUN T.
MEMG 13, 26 - 30 Ekim 2016
- XX. **a rare lysosomal storage disease**
COŞKUN T., canoruç d., BİLGİNER GÜRBÜZ B., PEKTAŞ E., DURSUN A., SİVRİ H. S., TOKATLI A.
MEMG 13, 26 - 30 Ekim 2016
- XXI. **Presentation of classic galactosemia with positive newborn screening for hyperphenylalaninemia**
DURSUN A., YILDIZ Y., akın s., bozat a., YIĞİT Ş., BİLGİNER GÜRBÜZ B., PEKTAŞ E., SİVRİ H. S., TOKATLI A., COŞKUN T.
13th Middle East Metabolic Group Meeting, 28 - 30 Ekim 2016
- XXII. **gnAL gene mutation and dystonia in two türkish siblings diagnosed by exom sequencing**
GÜNDÜZ M., ÜNAL Ö., ÖZGÜL R. K., DURSUN A.
SSIEM ROMA, 6 - 09 Eylül 2016
- XXIII. **Phenotypic variability and clinical biochemical histological andmolecular genetic characteristics of 17 patients with multipleacyl CoA dehydrogenase deficiency**
TOKATLI A., BİLGİNER GÜRBÜZ B., PEKTAŞ E., DURSUN A., SİVRİ H. S., COŞKUN T.
SSIEM ROMA, 6 - 09 Eylül 2016
- XXIV. **Prenatal findings and autopsy examination in a newborn with multipleacyl CoA dehydrogenase deficiency**
DOKUZBOY S., TALİM B., YIĞİT Ş., SİVRİ H. S., TOKATLI A., DURSUN A., COŞKUN T.
SSIEM ROMA, 6 - 09 Eylül 2016
- XXV. **Short term outcome of surgical correction of genu valgum in fourpatients with mucopolysaccharidosis type IV**
SİVRİ H. S., aksoy m., BİLGİNER GÜRBÜZ B., PEKTAŞ E., DURSUN A., TOKATLI A., COŞKUN T.
SSIEM RDMA, 6 Eylül - 09 Haziran 2016
- XXVI. **ethymelanonic encephopathy without etilmelanoc acitürria**
YÜCEL YILMAZ D., ÖZGÜL R. K., PEKTAŞ E., SERDARDĞLU e., YALNIZOĞLU D., DURSUN A.
SSIEM ROMA, 6 - 09 Eylül 2016
- XXVII. **the fist case of phenylketonuria with tyrosinemia type III**
COŞKUN T., BİLGİNER GÜRBÜZ B., DURSUN A., SİVRİ H. S., TOKATLI A.
SSIEM ROMA, 6 - 09 Eylül 2016
- XXVIII. **A case with psychomotor regression and leukoencephalopathy due to RNASEH2B gene defect**
ÖZGÜL R. K., YÜCEL YILMAZ D., SERDAROĞLU E., YALNIZOĞLU D., TOPÇU M., DURSUN A.
Annual Symposium of the Society for the Study of Inborn Errors of Metabolism (SSIEM), 6 - 09 Eylül 2016
- XXIX. **pontocerebellar hypoplasia type 6 a case with meonatal seizures hypotonia and microcephaly diagnosed by exome sequencing**
DURSUN A., sedaroğlu e., ÖZGÜL R. K., YÜCEL YILMAZ D., YALNIZOĞLU D.
SSIEM roma, 6 - 09 Eylül 2016
- XXX. **Ethylmalonic encephalopathy without ethylmalonic aciduria**
YÜCEL YILMAZ D., ÖZGÜL R. K., PEKTAŞ E., serdaroğlu e., YALNIZOĞLU D., DURSUN A.
Annual Symposium of the Society for the Study of Inborn Errors of Metabolism (SSIEM), 6 - 09 Eylül 2016
- XXXI. **argininosüccinic acidürria associated with pancreatitis**
DURSUN A., SİVRİ H. S., AKÇÖREN Z., TOKATLI A., COŞKUN T.
SSIEM, 6 - 09 Eylül 2016
- XXXII. **Adult mucopolysaccharidosis type VI patient with severe cervicalcord compression at diagnosis**
SİVRİ H. S., mocan ö., DURSUN A., TOKATLI A., COŞKUN T.
SSIEM roma, 6 - 09 Eylül 2016
- XXXIII. **Pontocerebellar hypoplasia type 6 a case with neonatal seizures hypotonia and microcephaly**

diagnosed by exome sequencing

SERDAROĞLU E., ÖZGÜL R. K., YÜCEL YILMAZ D., YALNIZOĞLU D., DURSUN A.

Annual Symposium of the Society for the Study of Inborn Errors of Metabolism (SSIEM), 6 - 09 Eylül 2016

XXXIV. Optic neuropathy a rare late complication in methylmalonicacidemia

BİLGİNER GÜRBÜZ B., PEKTAŞ E., DURSUN A., SİVRİ H. S., TOKATLI A., COŞKUN T.

SSIEM ROMA, 6 - 09 Eylül 2016

XXXV. Late diagnosed phenylketonuria in an eight year old boy with dyslexia and attention deficit

DURSUN A., TOKATLI A., COŞKUN T., SİVRİ H. S., YILDIZ Y.

SSIEM ROMA, 6 - 09 Eylül 2016

XXXVI. yeni metabolik hastalıkların tanımlanmasında genetik yaklaşım

DURSUN A.

V uluslararası katılımlı lizozomal hastalıklar kongresi, 14 - 17 Nisan 2016

XXXVII. Veziküler trafik bozuklukları

DURSUN A.

3. nörometabolik dismorfoloji sempozyumu, Türkiye, 10 - 13 Mart 2016

XXXVIII. Ekzom Dizi Analizi ile ATP8A2 Aminofosfolipid Transporter Protein Geninde Saptanan Yeni Bir Splaying Mutasyonu

YÜCEL YILMAZ D., DURSUN A., YALNIZOĞLU D., serdaroğlu e.

3. Nörometabolik Dismorfoloji Sempozyumu, Türkiye, 10 - 12 Mart 2016

XXXIX. Glutarik Asidemi Tip 2 Dismorfolojik İpuçları Veren Metabolik Bir Hastalık

AKGÜN DOĞAN Ö., ÜNSAL Y., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., DURSUN A., TALİM B., YİĞİT Ş.

3. Nörometabolik Dismorfoloji Sempozyumu, Türkiye, 10 - 12 Mart 2016

XL. Two cases with mucopolysaccharidosis type VII

Sivri S., Pektaş E., YILDIZ Y., DURSUN A., TOKATLI A., COŞKUN T.

12th Annual WORLD Symposium, California, Amerika Birleşik Devletleri, 29 Şubat - 04 Mart 2016, cilt.117

XLI. Türkiye de Genom Veri Bankasının Oluşturulması

DURSUN A.

7. DETAE günleri 2015, Türkiye, 11 - 12 Kasım 2015

XLII. 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 Eylül 2015

XLIII. Partial biotinidase deficiency with late onset severe cutaneous manifestations

SİVRİ H. S., yıldız Y., PEKTAŞ E., KK C., ALEHAN D., DURSUN A., TOKATLI A., COŞKUN T.

SSIEM, 4 - 07 Eylül 2015

XLIV. 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 Eylül 2015

XLV. Coexistence of phenylketonuria and primary adrenal insufficiency

COŞKUN T., PEKTAŞ E., GG b., DURSUN A., SİVRİ H. S., TOKATLI A.

SSIEM, 4 - 07 Eylül 2015

XLVI. 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 Eylül 2015

XLVII. Hyperlysinemia in a child and his mother

PEKTAŞ E., Burcu h., Özlem u., DURSUN A., SİVRİ H. S., TOKATLI A., COŞKUN T.

SSIEM, 4 - 07 Eylül 2015

XLVIII. A rare metabolic disease succinic semialdehyde dehydrogenase deficiency

TOKATLI A., PEKTAŞ E., yıldız Y., Özlem u., DURSUN A., SİVRİ H. S., COŞKUN T.

SSIEM, 4 - 07 Eylül 2015

XLIX. Two Cases with Mucopolysaccharidosis Type VII Sly s Syndrome

SİVRİ H. S., PEKTAŞ E., DURSUN A., TOKATLI A., COŞKUN T.

SSIEM, 4 - 07 Eylül 2015

- L. **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 Eylül 2015
- LI. **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 Eylül 2015
- LII. **Vacuolar storage material in a family with juvenile parkinsonism and mutations in FBX07**
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 Eylül 2015
- LIII. **Klasik Glutarik Asidüri Tip I GA I Bulguları Göstermeyen Bir Ailede GCDH Gen Defekti**
ÖZGÜL R. K., YÜCEL YILMAZ D., SANIYE Ö., YALNIZOĞLU D., TURANLI G., ESRA S., SİVRİ H. S., TOKATLI A., COŞKUN T., DURSUN A.
Uluslararası Katılımlı XIII.Ulusal Metabolik Hastalıklar Ve Beslenme Kongresi, Türkiye, 14 - 18 Nisan 2015
- LIV. **Türk İzovalerik Asidemi Hastalarında Genotip Fenotip İlişkisi**
Kılıç M., ÖZGÜL R. K., KARACA M., Küçük Ö., YÜCEL YILMAZ D., ALİFENDİOĞLU D., SİVRİ H. S., TOKATLI A., COŞKUN T., DURSUN A.
Uluslararası Katılımlı XIII.Ulusal Metabolik Hastalıklar Ve Beslenme Kongresi, Türkiye, 14 - 18 Nisan 2015
- LV. **Lizozom Otofagozom Defekti Sonucu Serebellar Atrofiye Neden Olan Yeni Bir Gen SNX14 Tanımlanması**
ÖZGÜL R. K., YÜCEL YILMAZ D., YALNIZOĞLU D., MAHMUT S., DURSUN A.
Uluslararası Katılımlı XIII.Ulusal Metabolik Hastalıklar Ve Beslenme Kongresi, 1, Türkiye, 14 - 18 Nisan 2015
- LVI. **Türk Hastalarda Biyotinidaz Gen Mutasyonlarının Moleküler Karakterizasyonu**
KARACA M., ÖZGÜL R. K., Ünal Ö., YÜCEL YILMAZ D., Kılıç M., Burcu H., TOKATLI A., COŞKUN T., DURSUN A., SİVRİ H. S.
Uluslararası Katılımlı XIII.Ulusal Metabolik Hastalıklar Ve Beslenme Kongresi, Türkiye, 14 - 18 Nisan 2015
- LVII. **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.
Uluslararası Katılımlı XIII.Ulusal Metabolik Hastalıklar Ve Beslenme Kongresi, Türkiye, 14 - 18 Nisan 2015
- LVIII. **Ekzom Dizi Analizi ile MTO1 Mitokondriyal tRNA Modifier Geninde Saptanan Yeni Bir Mutasyon**
YÜCEL YILMAZ D., ÖZGÜL R. K., ÖMER FARUK G., ESRA S., BETÜL Y., SAĞIROĞLU M., YALNIZOĞLU D., DURSUN A.
Uluslararası Katılımlı XIII.Ulusal Metabolik Hastalıklar Ve Beslenme Kongresi, Türkiye, 14 - 18 Nisan 2015
- LIX. **Fenilketonüride yenilikler**
DURSUN A.
Fenilketonüride yenilikler, Türkiye, 20 - 21 Şubat 2015
- LX. **bilinmeyen nörometabolik hastalıklara yaklaşım**
DURSUN A.
xiii ulusal metabolik hastalıklar ve beslenme kongresi, Türkiye, 14 - 18 Nisan 2015
- LXI. **Lecture Gathering data in databases for clinical purposes**
DURSUN A.
ESHG-PPPC Course Genetics in health care: Practice and Policies, 12 - 15 Şubat 2015
- LXII. **Two adult siblings with progressive walking difficulty and visual disturbances**
SİVRİ H. S., YILDIZ Y., Kiper P. O. S., DURSUN A., TOKATLI A., COŞKUN T.
11th Annual WORLD Symposium of the Lysosomal-Disease-Network, Florida, Amerika Birleşik Devletleri, 9 - 13 Şubat 2015, cilt.114
- LXIII. **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., Sağırođlu M., DURSUN A.
SSIEM, 4 - 07 Eylül 2015

- LXIV. **AUDIOLOGICAL OUTCOMES OF MPS II: BEFORE AND AFTER ENZYME REPLACEMENT THERAPY**
YİÇİT Ö., ÜNAL Ö., GENÇ G. A., HİŞMİ B., DURSUN A., TOKATLI A., COŞKUN T., SİVRİ H. S.
JOURNAL OF INHERITED METABOLIC DISEASE, 20 Ekim 2012, cilt.35, ss.145
- LXV. **Üre döngüsü bozukluklarına ikincil neonatal hiperamonemik koma tedavisinde ammonul sodyum benzoat ve sodyum fenilasetat kullanımı**
ÜNAL Ö., HİŞMİ B., SÜRMEİ ONAY Ö., TOKATLI A., SİVRİ H. S., DURSUN A., YİÇİT Ş., COŞKUN T.
20. Ulusal Neonatoloji Kongresi (UNEKO-20), Türkiye, 15 - 18 Nisan 2012
- LXVI. **A novel mutation in DGUOK gene in a Turkish newborn**
KILIÇ M., DURSUN A., SİVRİ H. S., TOKATLI A., AKÇÖREN Z., YİÇİT Ş., VEZİR E., SENECA S., LINDA D. M., COŞKUN T.
J Inherit Metab Dis 2010;33(1):S81, P-229. (SSIEM, Annual Symposium, 2010, İstanbul, Turkey), İstanbul, Türkiye,
31 Ağustos 2010
- LXVII. **Sialik Asit Depo Hastalığının Belirlenmesinde Nükleer Manyetik Rezonans Spektroskopisi**
UZGÖREN BARAN A., AKBAY N., DURSUN A., COŞKUN T., İMAMOĞLU Y.
Metabolizmanın Regülasyonu ve Metabolik Bozukluklar Lisansüstü Yaz Okulu, Trabzon, Türkiye, 29 Haziran - 06
Temmuz 2008

Desteklenen Projeler

- DURSUN A., UFUK 2020 Projesi, Identification of Molecular Pathology of Undiagnosed Patients with Mitochondrial Disorders by Whole Exome Sequencing (project number is BBMRI_03.), 2017 - Devam Ediyor
- TOKATLI A., DURSUN A., SİVRİ H. S., LAY İ., ÖZGÜL R. K., YÜCEL YILMAZ D., GÜLBAKAN B., YILDIZ Y., Yükseköğretim Kurumları Destekli Proje, HACETTEPE ÜNİVERSİTESİ İLERİ METABOLİK TESTLER LABORATUVARI ALTYAPISININ YENİLENMESİ, 2020 - 2024
- DURSUN A., Özaydın E., GÜLBAKAN B., ÖZGÜL R. K., Yükseköğretim Kurumları Destekli Proje, Galaktozemi ve Akçağaç Şurubu Hastalarında İnflamasyon İlişkinin Hedefsiz Metabolomik İle Araştırılması, 2019 - 2022
- DURSUN A., ÖZGÜL R. K., YÜCEL YILMAZ D., Yükseköğretim Kurumları Destekli Proje, MBOAT7 Gen Defektinin Protein İfadesinin Araştırılması, 2019 - 2022
- DURSUN A., YALNIZOĞLU D., YÜCEL YILMAZ D., ÖZGÜL R. K., Yükseköğretim Kurumları Destekli Proje, TANI KONULAMAYAN METABOLİK/NÖROMETABOLİK HASTALIKLARDA VEZİKÜLER TRAFİK BOZUKLUKLARININ ARAŞTIRILMASI, 2017 - 2021
- ÖZGÜL R. K., SÜMER E. S., DURSUN A., VARDAR ACAR N., Yükseköğretim Kurumları Destekli Proje, Metabolik Hastalıklarda Mitokondriyal Disfonksiyon ve Oksidatif Stres İlişkinin Araştırılması, 2019 - 2020
- DURSUN A., Yükseköğretim Kurumları Destekli Proje, Metabolik miyopati kursu, 2016 - 2018
- DURSUN A., Yükseköğretim Kurumları Destekli Proje, Sistasyonin Beta- Sentaz Eksikliğine Bağlı Homosistinüri Hastalarında Genotip- Fenotip İlişkinin Araştırılması, 2013 - 2016
- DURSUN A., Yükseköğretim Kurumları Destekli Proje, Ailesel Hiperkolesterolemili Çocuklarda Mutasyon Taraması, 2010 - 2016

Metrikler

Yayın: 196

Atıf (WoS): 544

Atıf (Scopus): 649

H-İndeks (WoS): 14

H-İndeks (Scopus): 16

Akademi Dışı Deneyim

Tunceli Devlet Hastanesi

Nevşehir, Kozaklı Merkez Sağlık Ocağı