

Prof. ALİ DURSUN

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International Researcher IDs

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Education Information

Expertise In Medicine, Hacettepe University, Tıp Fakültesi, Çocuk Sağlığı Hastalıkları Anabilim Dalı, Turkey 1988 - 1992

Dissertations

Doctorate, 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

Research Areas

Medicine, Health Sciences, Internal Medicine Sciences, Child Health and Diseases, Pediatric Endocrinology and Metabolism

Academic Titles / Tasks

Professor, Hacettepe University, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2010 - Continues

Associate Professor, Hacettepe University, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2005 - 2010

Assistant Professor, Hacettepe University, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 1998 - 2005

Lecturer PhD, Istanbul University, Health Sciences Institute, Genetik Anabilim Dalı, 1994 - 1998

Expert, Hacettepe University, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 1988 - 1992

Courses

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

Kalıtısal metabolik hastalıklar tanı ve izlem, Doctorate, 2017 - 2018, 2016 - 2017

malnütrsiyon, Undergraduate, 2017 - 2018

asidosis, Undergraduate, 2017 - 2018, 2016 - 2017

fenilketonüri, Undergraduate, 2017 - 2018, 2016 - 2017

Kalıtısal Metabolik hastalıkların moleküler temeli, Doctorate, 2017 - 2018, 2016 - 2017

yenidoğan taramalar, Undergraduate, 2017 - 2018, 2016 - 2017

dislipidemiler, Undergraduate, 2017 - 2018, 2016 - 2017

Published journal articles indexed by SCI, SSCI, and AHCI

- 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, vol.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, vol.147, no.1, pp.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, vol.140, no.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, vol.139, no.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, vol.35, no.11, pp.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, vol.179, pp.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, vol.64, no.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, vol.63, no.4, pp.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, vol.63, no.11, 2020 (SCI-Expanded)
- X. **Predictors of acute metabolic decompensation in children with maple syrup urine disease at the emergency department**
YILDIZ Y., Akcan Yildiz L., DURSUN A., TOKATLI A., COŞKUN T., TEKŞAM Ö., SİVRİ H. S.
EUROPEAN JOURNAL OF PEDIATRICS, vol.179, no.7, pp.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, vol.182, no.4, pp.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, vol.33, no.3, pp.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, vol.33, no.2, pp.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, vol.32, no.10, pp.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, vol.99, pp.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., Pektaş E., SİVRİ H. S., OĞUZ B., DURSUN A., TOKATLI A., COŞKUN T., HALİLOĞLU M.
EUROPEAN JOURNAL OF RADIOLOGY, vol.116, pp.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, vol.42, no.2, pp.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, vol.123, no.1, pp.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, vol.173, no.11, pp.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, vol.30, no.6, pp.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, vol.165, pp.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, vol.26, no.1, pp.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, vol.33, no.1, pp.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, vol.86, pp.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, vol.46, no.2, pp.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, vol.58, no.1, pp.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, pp.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, vol.174, no.8, pp.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, vol.9, no.3, pp.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, vol.47, no.5, pp.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, vol.57, pp.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., UNAL O., HISMI B., Aliefendioglu D., SIVRI S., TOKATLI A., et al.
EUROPEAN JOURNAL OF MEDICAL GENETICS, vol.57, no.10, pp.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, vol.34, no.3, pp.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, vol.534, no.2, pp.197-203, 2014 (SCI-Expanded)
- XXXVI. **Pregnancy and Lactation Outcomes in a Turkish Patient with Lysinuric Protein Intolerance**
Unal O., COŞKUN T., ORHAN D., Tokatl A., DURSUN A., ÖZTÜRK HİŞMİ B., ÖZYÜNCÜ Ö., Sivri S. H. K.
JIMD REPORTS, VOL 13, vol.13, pp.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, vol.28, no.11, pp.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, vol.28, no.11, pp.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, vol.55, no.6, pp.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, vol.58, no.10, pp.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, vol.521, no.2, pp.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, vol.9, pp.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, vol.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, vol.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, vol.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, vol.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, vol.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, vol.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, vol.35, 2012 (SCI-Expanded)
- L. **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, vol.35, 2012 (SCI-Expanded)
- LI. **Methylmalonic acidemia mimicking diabetic ketoacidosis in an infant**
Güven A., Cebeci N., DURSUN A., Aktekin E. H., Baumgartner M. R., Fowler B.
PEDIATRIC DIABETES, vol.13, no.6, 2012 (SCI-Expanded)
- LII. **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, vol.35, 2012 (SCI-Expanded)
- LIII. **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, vol.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, vol.54, no.4, pp.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, vol.46, no.3, pp.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, vol.2, pp.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, vol.3, pp.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, vol.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.
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- LX. OUTCOME OF ENZYME REPLACEMENT THERAPY IN PATIENTS WITH MUCOPOLYSACCHARIDOSIS TYPE VI**
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