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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
Kalıtsal 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
Kalıtsal 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**
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III. **Predictors of eventual requirement of phenylalanine-restricted diet in young infants with phenylalanine hydroxylase deficiency initially managed with sapropterin monotherapy**
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IV. **COVID-19 in inherited metabolic disorders: Clinical features and risk factors for disease severity**
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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.
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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**
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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.
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X. **Predictors of acute metabolic decompensation in children with maple syrup urine disease at the emergency department**
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XI. **Genotypes and estimated prevalence of phosphomannomutase 2 deficiency in Turkey differ significantly from those in Europe**
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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**
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XIII. **Presentation of 14 alkaptonuria patients from Turkey**
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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.
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XV. **Determinants of Riboflavin Responsiveness in Multiple Acyl-CoA Dehydrogenase Deficiency**
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XVI. **Clinical highlights of a very rare phospolipid remodeling disease due to MBOAT7 gene defect**
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XVII. **Imaging liver nodules in tyrosinemia type-1: A retrospective review of 16 cases in a tertiary pediatric hospital**
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CLINICAL DYSMORPHOLOGY, vol.26, no.1, pp.1-12, 2017 (SCI-Expanded)

XXIV. **Partial hydatidiform mole in a phenylketonuria patient treated with sapropterin dihydrochloride**
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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.
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XXVI. **Evaluation and identification of IDUA gene mutations in Turkish patients with mucopolysaccharidosis type I**
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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.
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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.
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XXX. **Ailevi Hiperkolesterolemili Hastaların Mutasyon Analiz Sonuçlarının Simone-Broome Kriterleriyle Değerlendirilmesi**
AYKAN H. H., ÖZGÜL R. K., Güzel A., COŞKUN T., DURSUN A.
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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.
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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.
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XXXVIII. **Vanishing White Matter With Hepatomegaly and Hypertriglyceridemia Attacks**
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XL. **Galactosemia in the Turkish population with a high frequency of Q188R mutation and distribution of**

Duarte-1 and Duarte-2 variations
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Guzel-Ozanturk A., ÖZGÜL R. K., Unal O., Hismi B., Aydin H. I., Sivri S., TOKATLI A., COŞKUN T., Aksoz E., DURSUN A.
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XLV. **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.
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Gokmen-Ozel H., Unal O., Kalkanoglu-Sivri H. S., Koksal G., DURSUN A., Tokatli A., Coskun T.
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L. **PROPIONIC ACIDEMIA PRESENTING WITH PERSISTENT PULMONARY HYPERTENSION IN TWO NEONATES**
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