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Kişisel Bilgiler

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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**
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- II. **TRAPPC6B biallelic variants cause a neurodevelopmental disorder with TRAPP II and trafficking disruptions**
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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**
KAHRAMAN A. B., YILDIZ Y., ÇIKI K., Erdal I., AKAR H. T., DURSUN A., TOKATLI A., SİVRİ S.
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- V. **Organic acidemias in the neonatal period: 30 years of experience in a referral center for inborn errors of metabolism**
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- VI. **An investigation of different intracellular parameters for Inborn Errors of Metabolism: Cellular stress, antioxidant response and autophagy**
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- VII. **Biallelic mutations in ELFN1 gene associated with developmental and epileptic encephalopathy and joint laxity**
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- VIII. **Clinical and molecular characteristics of carnitine-acylcarnitine translocase deficiency with c.270delC and a novel c.408C>A variant**
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- IX. **Glutaric aciduria type 1: Genetic and phenotypic spectrum in 53 patients**
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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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- 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**

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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 phospholipid 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**
ÖZCAN H. N., KARÇAALTINCABA M., Pektas E., SİVRİ H. S., OĞUZ B., DURSUN A., TOKATLI A., COŞKUN T., HALİLOĞLU M.
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- XVIII. **Expanding the phenotype of phospholipid remodelling disease due to MBOAT7 gene defect**
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- XIX. **The genotypic and phenotypic spectrum of MTO1 deficiency**
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- XX. **Genotypic-phenotypic features and enzyme replacement therapy outcome in patients with mucopolysaccharidosis VI from Turkey**
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- 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.
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- XXII. **Novel and prevalent CYP11B1 gene mutations in Turkish patients with 11-beta hydroxylase deficiency**
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- XXIII. **A probable new syndrome with the storage disease phenotype caused by the VPS33A gene mutation**
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- XXIV. **Partial hydatidiform mole in a phenylketonuria patient treated with sapropterin dihydrochloride**
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- 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**
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- XXVI. **Evaluation and identification of IDUA gene mutations in Turkish patients with mucopolysaccharidosis type I**
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- XXVII. **Late-diagnosed phenylketonuria in an eight-year-old boy with dyslexia and attention-deficit hyperactivity disorder**
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- XXVIII. **Sapropterin dihydrochloride treatment in Turkish hyperphenylalaninemic patients under age four**
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- XXIX. **Detection of biotinidase gene mutations in Turkish patients ascertained by newborn and family screening**
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- XXX. **Ailevi Hiperkolesterolemili Hastaların Mutasyon Analiz Sonuçlarının Simone-Broome Kriterleriyle Değerlendirilmesi**
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- XXXI. **Biallelic mutations in SNX14 cause a syndromic form of cerebellar atrophy and lysosome-autophagosome dysfunction**
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- XXXII. **Two Turkish siblings with MEGDEL syndrome due to novel SERAC1 genemutation.**
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- XXXIII. **Phenotypic and genotypic spectrum of Turkish patients with isovaleric acidemia**
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- 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.
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- 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**
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- XXXVII. **A Patient With Pyruvate Carboxylase Deficiency and Nemaline Rods on Muscle Biopsy**
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- XXXVIII. **Vanishing White Matter With Hepatomegaly and Hypertriglyceridemia Attacks**
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- XXXIX. **Cobalamin C defect: a patient of late-onset type with homozygous p. R132*mutation**
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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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- XLII. **Molecular and clinical evaluation of Turkish patients with lysinuric protein intolerance**
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- XLIII. **Novel Mutations in the PC Gene in Patients with Type B Pyruvate Carboxylase Deficiency**
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- XLIV. **ACUTE INTERMITTENT PORPHYRIA: STILL UNCALLED BY PHYSICIANS**
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- XLV. **MOLECULAR CHARACTERISATION OF BIOTINIDASE GENE MUTATIONS IN TURKISH PATIENTS; AN UPDATE OF THE RESULTS**
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- XLVI. **ENZYME REPLACEMENT THERAPY (ERT) RESULTS OF PATIENTS WITH MUCOPOLYSACCHARIDOSIS TYPE II (HUNTER SYNDROME)**
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- XLVII. **A PATIENT WITH PYRUVATE CARBOXYLASE DEFICIENCY AND NEMALINE RODS ON MUSCLE BIOPSY**
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- XLVIII. **DIETARY MANAGEMENT AND GROWTH OF TETRAHYDROBIOPTERIN RESPONSIVE TURKISH PKU PATIENTS**
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- XLIX. **OCTN2 GENE MUTATIONS IN TURKISH PATIENTS WITH PRIMARY CARNITINE DEFICIENCY**
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- L. **FEVER OF UNKNOWN ORIGIN IN A YOUNG ADULT WITH END-STAGE RENAL DISEASE, PREMATURE CORONARY ARTERY DISEASE AND POLYNEUROPATHY**
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- LI. **PROPIONIC ACIDEMIA PRESENTING WITH PERSISTENT PULMONARY HYPERTENSION IN TWO NEONATES**
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- LII. **Methylmalonic acidemia mimicking diabetic ketoacidosis in an infant**
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- LIII. **MOLYBDENUM COFACTOR (MOCO) DEFICIENCY TYPE B; CLINICAL, BIOCHEMICAL AND NEUROIMAGING FEATURES OF FIVE PATIENTS WITH TWO NOVEL MUTATIONS**
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- LIV. **SEVERE AZOTEMIA AND HYPERNATREMIC DEHYDRATION IN AN INFANT WITH PHENYLKETONURIA**
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- LIV. **Detection of other inborn errors of metabolism in hyperphenylalaninemic babies picked up on narrow-spectrum screening programs**

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- LV. Identification of a Novel Twinkle Mutation in a Family With Infantile Onset Spinocerebellar Ataxia by Whole Exome Sequencing**
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Atıf (WoS): 544

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Akademi Dışı Deneyim

Tunceli Devlet Hastanesi

Nevşehir, Kozaklı Merkez Sağlık Ocağı