

## **Prof.Dr. ALİ DURSUN**

### **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

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

malnütrsiyon, Lisans, 2017 - 2018

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

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### **SCI, SSCI ve AHCI İndekslerine Giren Dergilerde Yayınlanan Makaleler**

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- II. **TRAPP C6B biallelic variants cause a neurodevelopmental disorder with TRAPP II and trafficking disruptions**  
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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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- 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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- XII. **Oral health status of children with phenylketonuria**  
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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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- 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**

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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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- 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**  
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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**  
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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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- XLI. **Molecular and clinical evaluation of Turkish patients with lysinuric protein intolerance**  
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- XLII. **Novel Mutations in the PC Gene in Patients with Type B Pyruvate Carboxylase Deficiency**  
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- XLIII. **ACUTE INTERMITTENT PORPHYRIA: STILL UNCALLED BY PHYSICIANS**  
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- XLIV. **MOLECULAR CHARACTERISATION OF BIOTINIDASE GENE MUTATIONS IN TURKISH PATIENTS; AN UPDATE OF THE RESULTS**  
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- XLV. **ENZYME REPLACEMENT THERAPY (ERT)RESULTS OF PATIENTS WITH MUCOPOLYSACCHARIDOSIS TYPE II (HUNTER SYNDROME)**  
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- XLVI. **A PATIENT WITH PYRUVATE CARBOXYLASE DEFICIENCY AND NEMALINE RODS ON MUSCLE BIOPSY**  
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- LII. **MOLYBDENUM COFACTOR (MOCO) DEFICIENCY TYPE B; CLINICAL, BIOCHEMICAL AND NEUROIMAGING FEATURES OF FIVE PATIENTS WTH TWO NOVEL MUTATIONS**  
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- LIII. **SEVERE AZOTEMIA AND HYPERNATREMIC DEHYDRATION IN AN INFANT WITH PHENYLKETONURIA**  
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- IX. **Erişkinlerde kalıtsal metabolik hastalıklar: yatan hasta konsültasyonları ile üç yıllık deneyim**  
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- X. **Etildamelonik ensefalopati: vaka sunumu**  
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- XI. **Fenilketonürüli bireylerin diyetle fenilalanin ve protein alımları: Önerilere uyum nasıldır?**  
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- XXVII. **the fist case of phenylketonuria with tyrosinemai type III**  
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- XXIX. **Adult mucopolysaccharidosis type VI patient with severe cervicalcord compression at diagnosis**  
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- XXXIV. **Veziküler trafik bozuklukları**  
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- XXXVII. **Türkiye de Genom Veri Bankasının Oluşturulması**  
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7. DETAE günleri 2015, Türkiye, 11 - 12 Kasım 2015
- XXXVIII. **Rhizomelic chondrodysplasia punctata type II a case diagnosed by whole exome sequencing**  
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- XLV. **A rare metabolic disease succinic semialdehyde dehydrogenase deficiency**  
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- XLVI. **Mutation screening study in Turkish patients with L 2 hydroxyglutaric aciduria**  
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- XLVIII. **A case of fucosidosis with a new mutation in FUCA1 gene**  
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- XLIX. **Dirençli Hipoglisemi Hipertrofik Kardiyomiyopati ve Ensefalopatili Bir Hastada Ekzom Dizileme ile**

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**LI. bilinmeyen nörometabolik hastalıklara yaklaşım**

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**LIII. Two adult siblings with progressive walking difficulty and visual disturbances**

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## **Metrikler**

Yayın: 187

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