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### Eğitim Bilgileri

Tıpta Uzmanlık, Hacettepe Üniversitesi, Tıp Fakültesi, Çocuk Sağlığı Ve Hastalıkları, Türkiye 1993 - 1998

### Araştırma Alanları

Tıp, Sağlık Bilimleri, Dahili Tıp Bilimleri, Çocuk Sağlığı ve Hastalıkları

### Akademik Unvanlar / Görevler

Dr.Öğr.Üyesi, Hacettepe Üniversitesi, Tıp Fakültesi (ingilizce), Dahili Tıp Bilimleri Bölümü, 2018 - Devam Ediyor

Yrd.Doç.Dr., Hacettepe Üniversitesi, Tıp Fakültesi (ingilizce), Dahili Tıp Bilimleri Bölümü, 1999 - 2018

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

- I. **The Common miRNA Signatures Associated with Mitochondrial Dysfunction in Different Muscular Dystrophies**  
AKSU MENGEŞ E., Akkaya-Ulum Y. Z. , DAYANGAÇ ERDEN D., Balci-Peynircioglu B., YÜZBAŞIOĞLU A., Topaloglu H., TALİM B., Balci-Hayta B.  
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- II. **Circulating Epstein-Barr virus DNA and cell-free DNA in pediatric lymphomas**  
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- III. **Determinants of Riboflavin Responsiveness in Multiple Acyl-CoA Dehydrogenase Deficiency**  
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- IV. **Nemaline rod myopathy treated with L-tyrosine to relieve symptoms in a neonate**  
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- VI. **Bi-allelic mutations in MYL1 cause a severe congenital myopathy**  
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- VII. **LARGE expression in different types of muscular dystrophies other than dystroglycanopathy**  
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- VIII. **Prenatal Diagnosis of Merosin-Deficient Muscular Dystrophy**  
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- IX. **Non-immune hydrops fetalis: A retrospective analysis of 151 autopsies performed at a single center**  
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- X. **Clinical spectra of neuromuscular manifestations in patients with lipodystrophy: A multicenter study**  
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- XI. **Surgical Treatment of Childhood Inflammatory Myofibroblastic Tumors**  
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- XII. **Recessive PIEZO2 stop mutation causes distal arthrogryposis with distal muscle weakness, scoliosis and proprioception defects**  
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- XIII. **Congenital mirror movements in a patient with alpha-dystroglycanopathy due to a novel POMK mutation**  
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- XV. **Giant Omental Cyst (Lymphangioma) Mimicking Ascites and Tuberculosis**  
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- XVI. **Riboflavin-Responsive and -Non-responsive Mutations in FAD Synthase Cause Multiple Acyl-CoA Dehydrogenase and Combined Respiratory-Chain Deficiency**  
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- XVII. **Clinical characteristics of megaconial congenital muscular dystrophy due to choline kinase beta gene defects in a series of 15 patients**  
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- XX. **SPEG Interacts with Myotubularin, and Its Deficiency Causes Centronuclear Myopathy with Dilated Cardiomyopathy**  
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- XXI. **Mutation in TOR1AIP1 encoding LAP1B in a form of muscular dystrophy: A novel gene related to nuclear envelopathies**  
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- XXIII. **Unusual splenic hemangioma of a pediatric patient: hypointense on T2-weighted image**  
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- XXV. **Recessive TTN truncating mutations define novel forms of core myopathy with heart disease**  
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- XXVII. **A novel desmin mutation leading to autosomal recessive limb-girdle muscular dystrophy: distinct histopathological outcomes compared with desminopathies**  
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- XXVIII. **Mutations in KLHL40 Are a Frequent Cause of Severe Autosomal-Recessive Nemaline Myopathy**  
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- XXX. **Serum alpha-fetoprotein levels in neonatal cholestasis**  
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- XXXIII. **Coexistence of two distinct intragenic dystrophin deletions in two maternal cousins with Duchenne Muscular Dystrophy**

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- XXXIV. **Rhabdomyosarcoma of the common bile duct: an unusual cause of obstructive jaundice in a child**  
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- XXXVIII. **Nuclear factors involved in mitochondrial translation cause a subgroup of combined respiratory chain deficiency**  
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- XL. **Long-term follow-up in patients with congenital myasthenic syndrome due to CHAT mutations**  
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- XLI. **Double heterotopic pancreas and Meckel's diverticulum in a child: do they have a common origin?**  
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- XLII. **A RARE TYPE OF RENAL CELL CARCINOMA IN A GIRL: Hybrid Renal Cell Carcinoma**  
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- XLIII. **Sarcoid-like granulomas in common variable immunodeficiency**  
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- XLIV. **An unusual presentation of Muscle-Eye-Brain disease: Severe eye abnormalities with mild muscle and brain involvement**  
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- XLV. **A Comparative Study of alpha-Dystroglycan Glycosylation in Dystroglycanopathies Suggests that the Hypoglycosylation of alpha-Dystroglycan Does Not Consistently Correlate with Clinical Severity**  
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- XLVI. **Does Defective Apoptosis Play A Role in Cystic Fibrosis Lung Disease?**  
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- XLVII. **Ulcerative colitis associated with Takayasu's arteritis in a child**  
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**XLIX. Identification of a founder mutation in TPM3 in nemaline myopathy patients of Turkish origin**

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**LI. Pseudo-trisomy 13 in a fetus: further support for autosomal recessive inheritance**

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**LV. Histological response to injected dextranomer-based implant in a rat model**

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## Diğer Dergilerde Yayınlanan Makaleler

- I. **A Case of Acute Bilateral Ptosis: SURF-1 Mutation**  
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## Hakemli Kongre / Sempozyum Bildiri Kitaplarında Yer Alan Yayınlar

- I. **The profile of Duchenne muscular dystrophy patients younger than 10 years old from KUKAS registry, Turkey**  
KARADUMAN A. A. , GÜRBÜZ İ., ELİF A. A. , MEHMET G., BULUT N., AYDIN G., YILMAZ Ö., TALİM B., TOPALOĞLU H. A.  
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