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Education Information

Expertise In Medicine, Hacettepe Üniversitesi, Tıp Fakültesi, Çocuk Sağlığı Ve Hastalıkları, Turkey 1993 - 1998

Research Areas

Medicine, Health Sciences, Internal Medicine Sciences, Child Health and Diseases

Academic Titles / Tasks

Assistant Professor, Hacettepe Üniversitesi, Tıp Fakültesi (ingilizce), Dahili Tıp Bilimleri Bölümü, 2018 - Continues

Assistant Professor, Hacettepe Üniversitesi, Tıp Fakültesi (ingilizce), Dahili Tıp Bilimleri Bölümü, 1999 - 2018

Articles Published in Journals That Entered SCI, SSCI and AHCI Indexes

- 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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- 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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- V. **Do not Miss Rare and Treatable Cause of Early-Onset Hemolytic Uremic Syndrome: Cobalamin C Deficiency**
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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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- XIV. **Variants in the Oxidoreductase PYROXD1 Cause Early-Onset Myopathy with Internalized Nuclei and Myofibrillar Disorganization**
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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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- XVIII. **Next generation sequencing in a large cohort of patients presenting with neuromuscular disease before or at birth**
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- XIX. **Promotion of experimental autoimmune encephalomyelitis upon neutrophil granulocytes' stimulation with formyl-methionyl-leucyl-phenylalanine (fMLP) peptide**
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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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- XXII. **Use of Whole-Exome Sequencing to Determine the Genetic Basis of Multiple Mitochondrial Respiratory Chain Complex Deficiencies**
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- XXIV. **Vesiculopustular Eruption in Neonatal Transient Myeloproliferative Disorder**
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- XXV. **Recessive TTN truncating mutations define novel forms of core myopathy with heart disease**
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- XXVI. **Prenatal diagnosis and clinicopathologic examination of a case with diastematomyelia**
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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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- XXXII. **Solid tumors in Turkish children: a multicenter study**
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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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- XXXV. **Early corticosteroid treatment in 4 duchenne muscular dystrophy patients: 14-year follow-up**
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- XXXVI. **Colchicine Protects against Hyperoxic Lung Injury in Neonatal Rats**
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- XXXVIII. **Nuclear factors involved in mitochondrial translation cause a subgroup of combined respiratory chain deficiency**
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- XXXIX. **Mutation in Exon 1f of PLEC, Leading to Disruption of Plectin Isoform 1f, Causes Autosomal-Recessive Limb-Girdle Muscular Dystrophy**
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- XL. **Long-term follow-up in patients with congenital myasthenic syndrome due to CHAT mutations**
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- XLII. **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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XLVIII. Merkel Cell Carcinoma in a Child

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LIV. The myopathic form of coenzyme Q10 deficiency is caused by mutations in the electron-transferring-flavoprotein dehydrogenase (ETFDH) gene

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- II. **JÜVENİL DERMATOMİYOZİT HASTALARININ KAS BİYOPSİLERİNDE YARDIMCI T HÜCRE PROFİLLERİ**
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- III. **Inflammatory Pseudotumor: T Helper Cell Subtypes And Relation To IgG4-Related Disease?**
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- IV. **OC-7 Cresentic glomerulonephritis (CGN) in childhood classification of aetiology and clinicopathological importance of cd163 positive (M2) macrophages**
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- V. **The profile and natural history of congenital muscular dystrophies**
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- VIII. **Formyl-methionyl-leucyl-phenylalanine (fMLP) peptide as an adjuvant in experimental autoimmune encephalomyelitis (EAE): modulation of neutrophil granulocytes**
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