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Education Information

Expertise In Medicine, Hacettepe University, 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 University, Tıp Fakültesi (İngilizce), Dahili Tıp Bilimleri Bölümü, 2018 - Continues

Assistant Professor, Hacettepe University, Tıp Fakültesi (İngilizce), Dahili Tıp Bilimleri Bölümü, 1999 - 2018

Published journal articles indexed by SCI, SSCI, and AHCI

- I. **Early-onset juvenile dermatomyositis: A tertiary referral center experience and review of the literature**
ŞENER S., BAŞARAN H. Ö., BATU AKAL E. D., Sag E., Oz S., TALİM B., BİLGİNER Y., Halilolu G., ÖZEN S.
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- II. **A Child with Refractory and Relapsing Anti-3-Hydroxy-3-Methylglutaryl-Coenzyme A Reductase Myopathy: Case-Based Review**
ŞENER S., BATU AKAL E. D., SARI S., KASAP CÜCEOĞLU M., YILDIZ A. E., TALİM B., AYDINGÖZ Ü., ÖZEN S., Halilolu G.
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- III. **Alterations in insulin-like growth factor system in spinal muscular atrophy**
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- IV. **Could TGFBI-related corneal dystrophies be mimicked in zebrafish via CRISPR/Cas9-mediated hot spot arginine variations?**
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- V. **A Rare Pediatric Case of Severe Rhabdomyolysis Owing to Dual Infection**

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- VI. **A novel bi-allelic variant in the SDHB gene causes a severe mitochondrial complex II deficiency: a case report**
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- VII. **Reduced mitochondrial fission and impaired energy metabolism in human primary skeletal muscle cells of Megaconial Congenital Muscular Dystrophy**
AKSU MENGEŞ E., Eylem C. C., NEMUTLU E., Gizer M., KORKUSUZ P., Topaloglu H., TALİM B., Balci-Hayta B.
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- VIII. **Diagnostic yield of muscle biopsy in infants: Retrospective analysis of clinical and histopathologic findings**
Genc H. M., Guven A., TALİM B.
CLINICAL NEUROPATHOLOGY, vol.40, no.5, pp.286-291, 2021 (SCI-Expanded)
- IX. **Loss of C2orf69 defines a fatal autoinflammatory syndrome in humans and zebrafish that evokes a glycogen-storage-associated mitochondriopathy**
Wong H. H., Seet S. H., Maier M., GÜREL A., Traspas R. M., Lee C., Zhang S., TALİM B., Loh A. Y. T., Chia C. Y., et al.
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- X. **Milk of calcium: A rare manifestation of juvenile dermatomyositis**
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- XI. **Clinical and genetic characterization of PYROXD1-related myopathy patients from Turkey**
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- XII. **The Common miRNA Signatures Associated with Mitochondrial Dysfunction in Different Muscular Dystrophies**
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- XIII. **Circulating Epstein-Barr virus DNA and cell-free DNA in pediatric lymphomas**
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- XIV. **Determinants of Riboflavin Responsiveness in Multiple Acyl-CoA Dehydrogenase Deficiency**
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- XV. **Nemaline rod myopathy treated with L-tyrosine to relieve symptoms in a neonate**
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- XVI. **Do not Miss Rare and Treatable Cause of Early-Onset Hemolytic Uremic Syndrome: Cobalamin C Deficiency**
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- XVII. **LARGE expression in different types of muscular dystrophies other than dystroglycanopathy**
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- XVIII. **Bi-allelic mutations in MYL1 cause a severe congenital myopathy**
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HUMAN MOLECULAR GENETICS, vol.27, no.24, pp.4263-4272, 2018 (SCI-Expanded)
- XIX. **Prenatal Diagnosis of Merosin-Deficient Muscular Dystrophy**

- Fadiloglu E., Ozten G., Unal C., Talim B., Topaloglu H., Beksac M. S.
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- XX. **Non-immune hydrops fetalis: A retrospective analysis of 151 autopsies performed at a single center**
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- XXIII. **Recessive PIEZO2 stop mutation causes distal arthrogryposis with distal muscle weakness, scoliosis and proprioception defects**
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- XXIV. **Congenital mirror movements in a patient with alpha-dystroglycanopathy due to a novel POMK mutation**
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- XXVII. **Riboflavin-Responsive and -Non-responsive Mutations in FAD Synthase Cause Multiple Acyl-CoA Dehydrogenase and Combined Respiratory-Chain Deficiency**
Olsen R. K. J., Konarikova E., Giancaspero T. A., Mosegaard S., Boczonadi V., Matakovic L., Veauville-Merllie A., Terrile C., Schwarzmayr T., Haack T. B., et al.
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- XXVIII. **Next generation sequencing in a large cohort of patients presenting with neuromuscular disease before or at birth**
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- XXIX. **Clinical characteristics of megaconial congenital muscular dystrophy due to choline kinase beta gene defects in a series of 15 patients**
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- XXX. **Promotion of experimental autoimmune encephalomyelitis upon neutrophil granulocytes' stimulation with formyl-methionyl-leucyl-phenylalanine (fMLP) peptide**
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- XXXII. **Use of Whole-Exome Sequencing to Determine the Genetic Basis of Multiple Mitochondrial Respiratory Chain Complex Deficiencies**
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- XXXIII. **Unusual splenic hemangioma of a pediatric patient: hypointense on T2-weighted image**
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- XXXIV. **Mutation in TOR1AIP1 encoding LAP1B in a form of muscular dystrophy: A novel gene related to nuclear envelopathies**
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- XXXV. **Recessive TTN truncating mutations define novel forms of core myopathy with heart disease**
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- XXXVI. **Vesiculopustular eruption in neonatal transient myeloproliferative disorder**
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- XXXVII. **Prenatal diagnosis and clinicopathologic examination of a case with diastematomyelia**
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- XXXVIII. **Mutations in KLHL40 Are a Frequent Cause of Severe Autosomal-Recessive Nemaline Myopathy**
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- XXXIX. **A novel desmin mutation leading to autosomal recessive limb-girdle muscular dystrophy: distinct histopathological outcomes compared with desminopathies.**
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- XL. **Apoptosis in subacute sclerosing panencephalitis: Possibility for treatment**
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- XLI. **Serum alpha-fetoprotein levels in neonatal cholestasis**
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- XLII. **Multisystem fatal infantile disease caused by a novel homozygous EARS2 mutation**
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- XLIII. **Solid tumors in Turkish children: a multicenter study**
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- XLIV. **Coexistence of two distinct intragenic dystrophin deletions in two maternal cousins with Duchenne Muscular Dystrophy**
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- XLV. **Rhabdomyosarcoma of the common bile duct: an unusual cause of obstructive jaundice in a child**
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- XLVIII. **BRIEF REPORT A Dystroglycan Mutation Associated with Limb-Girdle Muscular Dystrophy**
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- LV. **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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- LVII. **An unusual presentation of Muscle-Eye-Brain disease: Severe eye abnormalities with mild muscle and brain involvement**
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- LVIII. **Ulcerative colitis associated with Takayasu's arteritis in a child**
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- LXV. **The myopathic form of coenzyme Q10 deficiency is caused by mutations in the electron-transferring-flavoprotein dehydrogenase (ETFDH) gene**
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- LXVI. **Histological response to injected dextranomer-based implant in a rat model**
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- LXVII. **Extranodal type T/NK-cell lymphoma with an atypical clinical presentation**
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- LXIX. **Escobar syndrome is a prenatal myasthenia caused by disruption of the acetylcholine receptor fetal gamma subunit**
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