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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. **Biallelic truncating variants in PACSIN3 cause childhood-onset myopathy with hyperCKaemia**
Distelmaier F., Sezer A., Helm C., Waldmueller S., Seibt A., Gangfuss A., Koelbel H., Schara-Schmidt U., Yuksel D., TALİM B., et al.
BRAIN, no.7, 2024 (SCI-Expanded)
- II. **Mimicking TGFBI Hot-Spot Mutation Did Not Result in Any Deposit Formation in the Zebrafish Cornea**
Yaylacioğlu Tuncay F., TALİM B., DİNÇER P. R.
Current Eye Research, vol.49, no.5, pp.458-466, 2024 (SCI-Expanded)
- III. **Early-onset juvenile dermatomyositis: A tertiary referral center experience and review of the literature**
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- IV. **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., Haliloglu G.
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- V. **Alterations in insulin-like growth factor system in spinal muscular atrophy**
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- VI. **Could TGFBI-related corneal dystrophies be mimicked in zebrafish via CRISPR/Cas9-mediated hot spot arginine variations?**
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EUROPEAN JOURNAL OF HUMAN GENETICS, vol.30, no.SUPPL 1, pp.140, 2022 (SCI-Expanded)
- VII. **A Rare Pediatric Case of Severe Rhabdomyolysis Owing to Dual Infection**
Goktas O. A., BEKTAŞ Ö., Tuncer G. O., ÖZÇAKAR Z. B., TALİM B., EMİNOĞLU F. T., TEBER S.
KLINISCHE PADIATRİE, vol.234, no.02, pp.119-122, 2022 (SCI-Expanded)
- VIII. **A novel bi-allelic variant in the SDHB gene causes a severe mitochondrial complex II deficiency: a case report**
PARILTAY E., PARILTAY E., TALİM B., Onay H.
CLINICAL NEUROLOGY AND NEUROSURGERY, vol.212, 2022 (SCI-Expanded)
- IX. **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)
- X. **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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- XI. **Loss of C2orf69 defines a fatal autoinflammatory syndrome in humans and zebrafish that evokes a glycogen-storage-associated mitochondriopathy**
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- XII. **Clinical and genetic characterization of PYROXD1-related myopathy patients from Turkey**
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- XIII. **Milk of calcium: A rare manifestation of juvenile dermatomyositis**
Yildirim D. G., AKDULUM İ., TALİM B., DEMİR E., BUYAN N., BAKKALOĞLU EZGÜ S. A.
ARCHIVES OF RHEUMATOLOGY, vol.36, no.2, pp.302-304, 2021 (SCI-Expanded)
- XIV. **Validation of the EULAR/ACR 2017 idiopathic inflammatory myopathy classification criteria in juvenile dermatomyositis patients.**
Sag E., Demir S., Bilginer Y., Talim B., Haliloglu G., Ozen S.
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- XV. **Knockout of zebrafish desmin genes does not cause skeletal muscle degeneration but alters calcium flux.**
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- XVII. **Inflammatory milieu of muscle biopsies in juvenile dermatomyositis.**
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- XVIII. **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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- XIX. **Circulating Epstein-Barr virus DNA and cell-free DNA in pediatric lymphomas**
YALÇIN B., Kutluk T., Agbaba S. K., Demir C., TALİM B.
TURKISH JOURNAL OF PEDIATRICS, vol.62, no.4, pp.541-550, 2020 (SCI-Expanded)
- XX. **Determinants of Riboflavin Responsiveness in Multiple Acyl-CoA Dehydrogenase Deficiency**
YILDIZ Y., TALİM B., Haliloglu G., Topaloglu H., AKÇÖREN Z., DURSUN A., SİVRİ H. S., COŞKUN T., TOKATLI A.
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- XXI. **Nemaline rod myopathy treated with L-tyrosine to relieve symptoms in a neonate**
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- XXII. **Do not Miss Rare and Treatable Cause of Early-Onset Hemolytic Uremic Syndrome: Cobalamin C Deficiency**
TOPALOĞLU R., Inozu M., GÜLHAN B., BİLGİNER GÜRBÜZ B., TALİM B., COŞKUN T.
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- XXIII. **LARGE expression in different types of muscular dystrophies other than dystroglycanopathy**
Balci-Hayta B., TALİM B., KALE G., Dincer P.
BMC NEUROLOGY, vol.18, 2018 (SCI-Expanded)
- XXIV. **Bi-allelic mutations in MYL1 cause a severe congenital myopathy**
Ravenscroft G., Zaharieva I. T., Bortolotti C. A., Lambrughi M., Pignataro M., Borsari M., Sewry C. A., Phadke R., Haliloglu G., Ong R., et al.
HUMAN MOLECULAR GENETICS, vol.27, no.24, pp.4263-4272, 2018 (SCI-Expanded)
- XXV. **Prenatal Diagnosis of Merosin-Deficient Muscular Dystrophy**
Fadiloglu E., Ozten G., Unal C., Talim B., Topaloglu H., Beksac M. S.
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- XXVI. **Clinical spectra of neuromuscular manifestations in patients with lipodystrophy: A multicenter study**
Akinci G., Topaloglu H., Demir T., ERŞEN DANYELİ A., TALİM B., Keskin F. E., Kadioglu P., Talip E., ALTAY C., YAYLALI G. F., et al.
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- XXVII. **Recessive PIEZO2 stop mutation causes distal arthrogryposis with distal muscle weakness, scoliosis and proprioception defects**
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- XXVIII. **Congenital mirror movements in a patient with alpha-dystroglycanopathy due to a novel POMK mutation**
Ardicli D., GÖÇMEN R., TALİM B., SPRUTE R., Haliloglu G., ÇIRAK S., Topaloglu H.
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- XXIX. **Variants in the Oxidoreductase PYROXD1 Cause Early-Onset Myopathy with Internalized Nuclei and Myofibrillar Disorganization**
O'Grady G. L., Best H. A., Sztal T. E., Schartner V., Sanjuan-Vazquez T., Donkervoort S., Neto O. A., Sutton R. B., Ilkovski B., Romero N. B., et al.
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- XXX. **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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- XXXI. **Next generation sequencing in a large cohort of patients presenting with neuromuscular disease before or at birth**

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- XXXII. **Clinical characteristics of megaconial congenital muscular dystrophy due to choline kinase beta gene defects in a series of 15 patients**
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- XXXIII. **Promotion of experimental autoimmune encephalomyelitis upon neutrophil granulocytes' stimulation with formyl-methionyl-leucyl-phenylalanine (fMLP) peptide**
KILIÇ A., ESENDAĞLI G., SAYAT G., TALİM B., KARABUDAK R., KURNE A.
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- XXXIV. **SPEG Interacts with Myotubularin, and Its Deficiency Causes Centronuclear Myopathy with Dilated Cardiomyopathy**
Agrawal P. B., Pierson C. R., Joshi M., Liu X., Ravenscroft G., Moghadaszadeh B., Talabere T., Viola M., Swanson L. C., Haliloglu G., et al.
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- XXXV. **Mutation in TOR1AIP1 encoding LAP1B in a form of muscular dystrophy: A novel gene related to nuclear envelopathies**
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- XXXVI. **Use of Whole-Exome Sequencing to Determine the Genetic Basis of Multiple Mitochondrial Respiratory Chain Complex Deficiencies**
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- XXXVII. **Recessive TTN truncating mutations define novel forms of core myopathy with heart disease**
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- XXXVIII. **Mutations in KLHL40 Are a Frequent Cause of Severe Autosomal-Recessive Nemaline Myopathy**
Ravenscroft G., Miyatake S., Lehtokari V., Todd E. J., Vomauen P., Yau K. S., Hayashi Y. K., Miyake N., Tsurusaki Y., Doi H., et al.
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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. **Multisystem fatal infantile disease caused by a novel homozygous EARS2 mutation**
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- XLII. **Coexistence of two distinct intragenic dystrophin deletions in two maternal cousins with Duchenne Muscular Dystrophy**
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- XLIII. **Early corticosteroid treatment in 4 duchenne muscular dystrophy patients: 14-year follow-up**
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- XLIV. **BRIEF REPORT A Dystroglycan Mutation Associated with Limb-Girdle Muscular Dystrophy**
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- XLV. **Nuclear factors involved in mitochondrial translation cause a subgroup of combined respiratory chain deficiency**
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- XLVI. **Mutation in exon 1f of PLEC, leading to disruption of plectin isoform 1f, causes autosomal-recessive limb-girdle muscular dystrophy.**
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- XLVII. **An unusual presentation of Muscle-Eye-Brain disease: Severe eye abnormalities with mild muscle and brain involvement**
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- XLVIII. **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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- XLIX. **Identification of a founder mutation in TPM3 in nemaline myopathy patients of Turkish origin**
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- L. **Lissencephaly type II**
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- LI. **The myopathic form of coenzyme Q10 deficiency is caused by mutations in the electron-transferring-flavoprotein dehydrogenase (ETFDH) gene**
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- LIII. **Escobar syndrome is a prenatal myasthenia caused by disruption of the acetylcholine receptor fetal gamma subunit**
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- LIV. **Calpain-3 mutations in Turkey**
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- LV. **Focal segmental glomerulosclerosis associated with mitochondrial cytopathy: Report of two cases with special emphasis on podocytes**
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- LVI. **Prenatal diagnosis in laminin alpha 2 chain (merosin)-deficient congenital muscular dystrophy: A collective experience of five international centers**

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- LVIII. **Myogenesis within the human gubernaculum: Histological and immunohistochemical evaluation**
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- LIX. **An autosomal recessive limb girdle muscular dystrophy (LGMD2) with mild mental retardation is allelic to Walker-Warburg syndrome (WWS) caused by a mutation in the POMT1 gene**
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- LX. **Severe lethal spinal muscular atrophy variant with arthrogryposis**
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- I. **A Case of Acute Bilateral Ptosis: SURF-1 Mutation**
 Dedeoglu O., Yuksel D., Sel C. G., Kilic M., Oguz K. K., TALİM B.
 TURKISH JOURNAL OF NEUROLOGY, vol.24, no.1, pp.83-85, 2018 (ESCI)
- II. **Beta-sarcoglycan gene mutations in Turkey.**
 Balci B., Wilichowski E., Haliloğlu G., Talim B., Aurino S., Kremer E., Ebinger F., Senbil N., Anlar B., Kale G., et al.
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Refereed Congress / Symposium Publications in Proceedings

- I. **Validation of the Euler/ACR 2017 idiopathic inflammatory myopathy classification criteria in JDM patients**
 SAĞ E., DEMİR S., BİLGİNER Y., TALİM B., HALILOĞLU V. G., ÖZEN S.
 Pediatric Rheumatology European Society (PREs) e-Congress-2020, BİLİNMEYEN ÜLKELER (DİĞER), 23 - 26 September 2020, vol.18, pp.54
- II. **Limb-Girdle Muscular Dystrophy 2R modelling in zebrafishto determine a novel mechanism related to desmin-lamin B interaction**
 KAYMAN KÜREKÇİ G., KURAL MANGIT E., ÜNSAL Ş., YERSAL N., ERGİN B., SAĞLAM B., DÜZ N., ÇINAR Z., TALİM B.,

KORKUSUZ P., et al.

Keystone Symposia, 11 - 15 November 2018

- III. **Disease Modeling in Zebrafish: Limb-Girdle Muscular Dystrophy 2R**
ÜNSAL Ş., KAYMAN KÜREKÇİ G., KURAL MANGIT E., TALİM B., YERSAL N., ERGİN B., DÜZ N., ÇINAR Z., KORKUSUZ P., PURALI N., et al.
6th International Congress of the Molecular Biology Association of Turkey, Turkey, 5 - 08 September 2018, pp.5-6
- IV. **JÜVENİL DERMATOMİYOZİT HASTALARININ KAS BİYOPSİLERİNDE YARDIMCI T HÜCRE PROFİLLERİ**
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Metrics

Publication: 146

Citation (WoS): 2546

Citation (Scopus): 2838

H-Index (WoS): 25

H-Index (Scopus): 26