

## Dr. Öğr. Üyesi BERİL TALİM

### Kişisel Bilgiler

**İş Telefonu:** [+90 312 305 1304](tel:+903123051304)

**Fax Telefonu:** [+90 312 305 1177](tel:+903123051177)

**E-posta:** btalim@hacettepe.edu.tr

**Web:** <https://avesis.hacettepe.edu.tr/btalim>

**Posta Adresi:** Hacettepe Çocuk Hastanesi, Patoloji Ünitesi 06100 Ankara

### Uluslararası Araştırmacı ID'leri

ORCID: 0000-0003-2375-7543

Yoksis Araştırmacı ID: 182279

### 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 (İngilizce), Dahili Tıp Bilimleri Bölümü, 2018 - Devam Ediyor  
Yrd. Doç. Dr., Hacettepe Üniversitesi, Tıp Fakültesi (İngilizce), Dahili Tıp Bilimleri Bölümü, 1999 - 2018

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

#### I. Anti-SRP myositis: a diagnostic and therapeutic challenge

Polat M. C., Ardicli D., Acar B. c., TALİM B., Senbil N., Celikel E.

TURKISH JOURNAL OF PEDIATRICS, sa.6, ss.792-800, 2024 (SCI-Expanded)

#### II. Six Years Follow-Up of an 11-Year-Old Girl with Anti-HMGCR Myopathy

Cavusoglu D., TALİM B., Ekinci G., Topaloglu H.

JOURNAL OF NEUROMUSCULAR DISEASES, cilt.11, sa.4, ss.883-887, 2024 (SCI-Expanded)

#### III. 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, cilt.147, sa.7, 2024 (SCI-Expanded)

#### IV. Mimicking TGFBI Hot-Spot Mutation Did Not Result in Any Deposit Formation in the Zebrafish Cornea

Yaylacioglu Tuncay F., TALİM B., DİNÇER P. R.

Current Eye Research, cilt.49, sa.5, ss.458-466, 2024 (SCI-Expanded)

#### V. 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., Haliloglu G., ÖZEN S.

- Seminars in Arthritis and Rheumatism, cilt.58, 2023 (SCI-Expanded)
- VI. **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.  
Journal of neuromuscular diseases, cilt.10, sa.2, ss.279-291, 2023 (SCI-Expanded)
- VII. **Alterations in insulin-like growth factor system in spinal muscular atrophy**  
YEŞBEK KAYMAZ A., Bal S. K., Bora G., TALİM B., Ozon A., ALİKAŞİFOĞLU A., Topaloglu H., YURTER H.  
MUSCLE & NERVE, cilt.66, sa.5, ss.631-638, 2022 (SCI-Expanded)
- VIII. **Could TGFBI-related corneal dystrophies be mimicked in zebrafish via CRISPR/Cas9-mediated hot spot arginine variations?**  
Tuncay F. Y., TALİM B., DİNÇER P. R.  
EUROPEAN JOURNAL OF HUMAN GENETICS, cilt.30, sa.SUPPL 1, ss.140, 2022 (SCI-Expanded)
- IX. **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 PEDIATRIE, cilt.234, sa.02, ss.119-122, 2022 (SCI-Expanded)
- X. **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, cilt.212, 2022 (SCI-Expanded)
- XI. **Diagnostic yield of muscle biopsy in infants: Retrospective analysis of clinical and histopathologic findings**  
Genc H. M., Guven A., TALİM B.  
CLINICAL NEUROPATHOLOGY, cilt.40, sa.5, ss.286-291, 2021 (SCI-Expanded)
- XII. **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.  
SCIENTIFIC REPORTS, cilt.11, sa.1, 2021 (SCI-Expanded)
- XIII. **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.  
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.108, sa.7, ss.1301-1317, 2021 (SCI-Expanded)
- XIV. **Milk of calcium: A rare manifestation of juvenile dermatomyositis**  
Yıldırım D. G., AKDULUM İ., TALİM B., DEMİR E., BUYAN N., BAKKALOĞLU EZGÜ S. A.  
ARCHIVES OF RHEUMATOLOGY, cilt.36, sa.2, ss.302-304, 2021 (SCI-Expanded)
- XV. **Clinical and genetic characterization of PYROXD1-related myopathy patients from Turkey**  
Daimagueuer H., Akpulat U., Oezdemir O., YİŞ U., GÜNGÖR S., TALİM B., Diniz G., Baydan F., Thiele H., Altmueller J., et al.  
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.185, sa.6, ss.1678-1690, 2021 (SCI-Expanded)
- XVI. **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.  
Clinical and experimental rheumatology, cilt.39, ss.688-694, 2021 (SCI-Expanded)
- XVII. **Knockout of zebrafish desmin genes does not cause skeletal muscle degeneration but alters calcium flux.**  
Kayman Kürekçi G., Kural Mangit E., Koyunlar C., Unsal S., Saglam B., Ergin B., Gizer M., Uyanik I., Boustanabadimaralan Düz N., Korkusuz P., et al.  
Scientific reports, cilt.11, sa.1, ss.7505, 2021 (SCI-Expanded)
- XVIII. **Clinical features, muscle biopsy scores, myositis specific antibody profiles and outcome in juvenile dermatomyositis.**  
Sag E., Demir S., Bilginer Y., Talim B., Haliloglu G., Topaloglu H., Ozen S.

- Seminars in arthritis and rheumatism, cilt.51, ss.95-100, 2020 (SCI-Expanded)
- XIX. **Inflammatory milieu of muscle biopsies in juvenile dermatomyositis.**  
Sag E., Kale G., Haliloglu G., Bilginer Y., Akcoren Z., Orhan D., Gucer S., Topaloglu H., Ozen S., Talim B.  
Rheumatology international, 2020 (SCI-Expanded)
- XX. **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.  
AMERICAN JOURNAL OF PATHOLOGY, cilt.190, sa.10, ss.2136-2145, 2020 (SCI-Expanded)
- XXI. **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, cilt.62, sa.4, ss.541-550, 2020 (SCI-Expanded)
- XXII. **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.  
PEDIATRIC NEUROLOGY, cilt.99, ss.69-75, 2019 (SCI-Expanded)
- XXIII. **Nemaline rod myopathy treated with L-tyrosine to relieve symptoms in a neonate**  
Sahin S., Oncel M. Y., Bidev D., Okur N., TALİM B., Oguz S. S.  
ARCHIVOS ARGENTINOS DE PEDIATRIA, cilt.117, sa.4, 2019 (SCI-Expanded)
- XXIV. **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.  
NEPHRON, cilt.142, sa.3, ss.258-263, 2019 (SCI-Expanded)
- XXV. **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, cilt.27, sa.24, ss.4263-4272, 2018 (SCI-Expanded)
- XXVI. **LARGE expression in different types of muscular dystrophies other than dystroglycanopathy**  
Balci-Hayta B., TALİM B., KALE G., Dincer P.  
BMC NEUROLOGY, cilt.18, 2018 (SCI-Expanded)
- XXVII. **Prenatal Diagnosis of Merosin-Deficient Muscular Dystrophy**  
Fadiloglu E., Ozten G., Unal C., Talim B., Topaloglu H., Beksac M. S.  
FETAL AND PEDIATRIC PATHOLOGY, cilt.37, sa.6, ss.418-423, 2018 (SCI-Expanded)
- XXVIII. **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.  
NEUROMUSCULAR DISORDERS, cilt.27, sa.10, ss.923-930, 2017 (SCI-Expanded)
- XXIX. **Recessive PIEZO2 stop mutation causes distal arthrogryposis with distal muscle weakness, scoliosis and proprioception defects**  
HALILOGLU G., BECKER K., Temucin C., TALİM B., KUCUKSAHIN N., PERGANDE M., MOTAMENY S., NURNBERG P., AYDINGÖZ Ü., TOPALOĞLU H., et al.  
JOURNAL OF HUMAN GENETICS, cilt.62, sa.4, ss.497-501, 2017 (SCI-Expanded)
- XXX. **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.  
NEUROMUSCULAR DISORDERS, cilt.27, sa.3, ss.239-242, 2017 (SCI-Expanded)
- XXXI. **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.  
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.99, sa.5, ss.1086-1105, 2016 (SCI-Expanded)
- XXXII. **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.  
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.98, sa.6, ss.1130-1145, 2016 (SCI-Expanded)
- XXXIII. Next generation sequencing in a large cohort of patients presenting with neuromuscular disease before or at birth**  
Todd E. J., Yau K. S., Ong R., Slee J., McGillivray G., Barnett C. P., Haliloglu G., TALİM B., AKÇÖREN Z., Kariminejad A., et al.  
ORPHANET JOURNAL OF RARE DISEASES, cilt.10, 2015 (SCI-Expanded)
- XXXIV. Clinical characteristics of megaonial congenital muscular dystrophy due to choline kinase beta gene defects in a series of 15 patients**  
Haliloglu G., TALİM B., Sel C. G., Topaloglu H.  
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.38, sa.6, ss.1099-1108, 2015 (SCI-Expanded)
- XXXV. 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.  
AUTOIMMUNITY, cilt.48, sa.6, ss.423-428, 2015 (SCI-Expanded)
- XXXVI. 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.  
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.95, sa.2, ss.218-226, 2014 (SCI-Expanded)
- XXXVII. Use of Whole-Exome Sequencing to Determine the Genetic Basis of Multiple Mitochondrial Respiratory Chain Complex Deficiencies**  
Taylor R. W., Pyle A., Griffin H., Blakely E. L., Duff J., He L., Smertenko T., Alston C. L., Neeve V. C., Best A., et al.  
JAMA-JOURNAL OF THE AMERICAN MEDICAL ASSOCIATION, cilt.312, sa.1, ss.68-77, 2014 (SCI-Expanded)
- XXXVIII. Mutation in TOR1AIP1 encoding LAP1B in a form of muscular dystrophy: A novel gene related to nuclear envelopathies**  
Kayman-Kurekci G., TALİM B., KORKUSUZ P., Sayar N., Sarioglu T., Oncel I., Sharafi P., Gundesli H., Balci-Hayta B., PURALI N., et al.  
NEUROMUSCULAR DISORDERS, cilt.24, sa.7, ss.624-633, 2014 (SCI-Expanded)
- XXXIX. Recessive TTN truncating mutations define novel forms of core myopathy with heart disease**  
Chauveau C., Bonnemann C. G., Julien C., Kho A. L., Marks H., TALİM B., Maury P., Arne-Bes M. C., Uro-Coste E., Alexandrovich A., et al.  
HUMAN MOLECULAR GENETICS, cilt.23, sa.4, ss.980-991, 2014 (SCI-Expanded)
- XL. A novel desmin mutation leading to autosomal recessive limb-girdle muscular dystrophy: distinct histopathological outcomes compared with desminopathies.**  
Cetin N., Balci-Hayta B., Gundesli H., KORKUSUZ P., PURALI N., TALİM B., Tan E., Selcen D., Erdem-Ozdamar S., Dincer P. R.  
Journal of medical genetics, cilt.50, sa.7, ss.437-43, 2013 (SCI-Expanded)
- XLI. 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.  
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.93, sa.1, ss.6-18, 2013 (SCI-Expanded)
- XLII. Apoptosis in subacute sclerosing panencephalitis: Possibility for treatment**  
Anlar B., BEKEN S., TALİM B.  
MEDICAL HYPOTHESES, cilt.80, sa.4, ss.507-508, 2013 (SCI-Expanded)
- XLIII. Multisystem fatal infantile disease caused by a novel homozygous EARS2 mutation**  
TALİM B., Pyle A., Griffin H., Topaloglu H., TOKATLI A., Keogh M. J., Santibanez-Koref M., Chinnery P. F., Horvath R.  
BRAIN, cilt.136, 2013 (SCI-Expanded)
- XLIV. Coexistence of two distinct intragenic dystrophin deletions in two maternal cousins with Duchenne**

**Muscular Dystrophy**

Balci-Hayta B., TALİM B., Dincer P., Topaloglu H.

NEUROMUSCULAR DISORDERS, cilt.23, sa.1, ss.15-18, 2013 (SCI-Expanded)

- XLV. **Early corticosteroid treatment in 4 duchenne muscular dystrophy patients: 14-year follow-up**  
Merlini L., Gennari M., Malaspina E., Cecconi I., Armaroli A., Gnudi S., TALİM B., Ferlini A., Cicognani A., Franzoni E.  
MUSCLE & NERVE, cilt.45, sa.6, ss.796-802, 2012 (SCI-Expanded)
- XLVI. **BRIEF REPORT A Dystroglycan Mutation Associated with Limb-Girdle Muscular Dystrophy**  
Hara Y., Balci-Hayta B., Yoshida-Moriguchi T., Kanagawa M., de Bernabe D. B., Gundesli H., Willer T., Satz J. S.,  
Crawford R. W., Burden S. J., et al.  
NEW ENGLAND JOURNAL OF MEDICINE, cilt.364, sa.10, ss.939-946, 2011 (SCI-Expanded)
- XLVII. **Nuclear factors involved in mitochondrial translation cause a subgroup of combined respiratory chain deficiency**  
Kemp J. P., Smith P. M., Pyle A., Neeve V. C. M., Tuppen H. A. L., Schara U., TALİM B., Topaloglu H., Holinski-Feder E.,  
Abicht A., et al.  
BRAIN, cilt.134, ss.183-195, 2011 (SCI-Expanded)
- XLVIII. **Mutation in exon 1f of PLEC, leading to disruption of plectin isoform 1f, causes autosomal-recessive limb-girdle muscular dystrophy.**  
Gundesli H., TALİM B., KORKUSUZ P., Balci-Hayta B., Cirak S., Akarsu N. A., Topaloglu H., Dincer P. R.  
American journal of human genetics, cilt.87, sa.6, ss.834-41, 2010 (SCI-Expanded)
- XLIX. **An unusual presentation of Muscle-Eye-Brain disease: Severe eye abnormalities with mild muscle and brain involvement**  
DEMİR E., GÜCÜYENER K., Akturk A., TALİM B., Konus O., Del Bo R., Ghezzi S., Comi G. P.  
NEUROMUSCULAR DISORDERS, cilt.19, sa.10, ss.692-695, 2009 (SCI-Expanded)
- L. **A Comparative Study of alpha-Dystroglycan Glycosylation in Dystroglycanopathies Suggests that the Hypoglycosylation of alpha-Dystroglycan Does Not Consistently Correlate with Clinical Severity**  
Jimenez-Mallebrera C., Torelli S., Feng L., Kim J., Godfrey C., Clement E., Mein R., Abbs S., Brown S. C., Campbell K. P.,  
et al.  
BRAIN PATHOLOGY, cilt.19, sa.4, ss.596-611, 2009 (SCI-Expanded)
- LI. **Identification of a founder mutation in TPM3 in nemaline myopathy patients of Turkish origin**  
Lehtokari V., Pelin K., Donner K., Voit T., Rudnik-Schoenborn S., Stoetter M., TALİM B., Topaloglu H., Laing N. G.,  
Wallgren-Pettersson C.  
EUROPEAN JOURNAL OF HUMAN GENETICS, cilt.16, sa.9, ss.1055-1061, 2008 (SCI-Expanded)
- LII. **Lissencephaly type II**  
Topaloglu H., TALİM B.  
MALFORMATIONS OF THE NERVOUS SYSTEM, cilt.87, ss.219-234, 2008 (SCI-Expanded)
- LIII. **The myopathic form of coenzyme Q10 deficiency is caused by mutations in the electron-transferring-flavoprotein dehydrogenase (ETFDH) gene**  
Gempel K., Topaloglu H., Talim B., Schneiderat P., Schoser B. G. H., Hans V. H., Palmafay B., Kale G., Tokatli A., Quinzii C., et al.  
BRAIN, cilt.130, ss.2037-2044, 2007 (SCI-Expanded)
- LIV. **Prenatal diagnosis of muscle-eye-brain disease**  
Balci B., Morris-Rosendahl D. J., Celebi A., Talim B., Topaloglu H., Dincer P. R.  
PRENATAL DIAGNOSIS, cilt.27, sa.1, ss.51-54, 2007 (SCI-Expanded)
- LV. **Escobar syndrome is a prenatal myasthenia caused by disruption of the acetylcholine receptor fetal gamma subunit**  
Hoffmann K., Mueller J. S., Stricker S., Megarbane A., Rajab A., Lindner T. H., Cohen M., Chouery E., Adaimy L.,  
Ghanem I., et al.  
AMERICAN JOURNAL OF HUMAN GENETICS, cilt.79, sa.2, ss.303-312, 2006 (SCI-Expanded)
- LVI. **Calpain-3 mutations in Turkey**  
Balci B., Aurino S., Haliloglu G., Talim B., Erdem S., Akcoren Z., Tan E., Caglar M., Richard I., Nigro V., et al.  
EUROPEAN JOURNAL OF PEDIATRICS, cilt.165, sa.5, ss.293-298, 2006 (SCI-Expanded)

- LVII. **Focal segmental glomerulosclerosis associated with mitochondrial cytopathy: Report of two cases with special emphasis on podocytes**  
 Gucer S., Talim B., Asan E., Korkusuz P., Ozen S., Unal S., Kalkanoglu S., Kale G., Caglar M.  
 PEDIATRIC AND DEVELOPMENTAL PATHOLOGY, cilt.8, sa.6, ss.710-717, 2005 (SCI-Expanded)
- LVIII. **Prenatal diagnosis in laminin alpha 2 chain (merosin)-deficient congenital muscular dystrophy: A collective experience of five international centers**  
 Vainzof M., Richard P., Herrmann R., Jimenez-Mallebrera C., Talim B., Yamamoto L., Ledeuil C., Mein R., Abbs S., Brockington M., et al.  
 NEUROMUSCULAR DISORDERS, cilt.15, ss.588-594, 2005 (SCI-Expanded)
- LIX. **Dominant and recessive COL6A1 mutations in Ullrich scleroatonic muscular dystrophy**  
 Giusti B., Lucarini L., Pietroni V., Lucioli S., Bandinelli B., Sabatelli P., Squarzoni S., Petrini S., Gartioux C., Talim B., et al.  
 ANNALS OF NEUROLOGY, cilt.58, sa.3, ss.400-410, 2005 (SCI-Expanded)
- LX. **Myogenesis within the human gubernaculum: Histological and immunohistochemical evaluation**  
 TANYEL F. C., TALIM B., ATILLA P., Muftuoglu S. F., KALE G.  
 EUROPEAN JOURNAL OF PEDIATRIC SURGERY, cilt.15, sa.3, ss.175-179, 2005 (SCI-Expanded)
- LXI. **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**  
 Balci B., Uyanik G., Dincer P. R., Gross C., Willer T., Talim B., Haliloglu G., Kale G., Hehr U., Winkler J., et al.  
 NEUROMUSCULAR DISORDERS, cilt.15, sa.4, ss.271-275, 2005 (SCI-Expanded)
- LXII. **Severe lethal spinal muscular atrophy variant with arthrogryposis**  
 Kizilates S., Talim B., Sel K., Kose G., Caglar M.  
 PEDIATRIC NEUROLOGY, cilt.32, sa.3, ss.201-204, 2005 (SCI-Expanded)
- LXIII. **A novel form of recessive limb girdle muscular dystrophy with mental retardation and abnormal expression of alpha-dystroglycan**  
 Dincer P. R., Balci B., Yuva Y., Talim B., Brockington M., Dincel D., Torelli S., Sue B., Kale G., Haliloglu G., et al.  
 NEUROMUSCULAR DISORDERS, cilt.13, sa.10, ss.771-778, 2003 (SCI-Expanded)
- LXIV. **Merosin-deficient congenital muscular dystrophy with mental retardation and cerebellar cysts unlinked to the LAMA2, FCMD and MEB loci**  
 Talim B., Ferreiro A., Cormand B., Vignier N., Oto A., Gogus S., Cila A., Lehesjoki A., Pihko H., Guicheney P., et al.  
 NEUROMUSCULAR DISORDERS, cilt.10, sa.8, ss.548-552, 2000 (SCI-Expanded)
- LXV. **Congenital epulis of the newborn - A case report**  
 Talim B., Yigit S., Oran O., Akcoren Z.  
 TURKISH JOURNAL OF PEDIATRICS, cilt.40, sa.1, ss.127-129, 1998 (SCI-Expanded)

## Diger Dergilerde Yayınlanan Makaleler

- I. **A Case of Acute Bilateral Ptosis: SURF-1 Mutation**  
 Dedeoglu O., Yuksel D., Sel C. G., Kilic M., Oguz K. K., TALIM B.  
 TURKISH JOURNAL OF NEUROLOGY, cilt.24, sa.1, ss.83-85, 2018 (ESCI)
- II. **Beta-sarcoglycan gene mutations in Turkey.**  
 Balci B., Wilichowski E., Haliloglu G., Talim B., Aurino S., Kremer E., Ebinger F., Senbil N., Anlar B., Kale G., et al.  
 Acta myologica : myopathies and cardiomyopathies : official journal of the Mediterranean Society of Myology, cilt.23, ss.154-8, 2004 (Scopus)

## Hakemli Kongre / Sempozyum Bildiri Kitaplarında Yer Alan Yayınlar

- I. **Validation of the Eular/ACR 2017 idiopathic inflammatory myopathy classification criteria in JDM patients**

- SAĞ E., DEMİR S., BİLGİNER Y., TALİM B., HALİLÖĞLU V. G., ÖZEN S.  
Pediatric Rheumatology European Society (PReS) e-Congress-2020, BİLİNMEYEN ÜLKELER (DİĞER), 23 - 26 Eylül 2020, cilt.18, ss.54
- II. Limb-Girdle Muscular Dystrophy 2R modelling in zebrafish to 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 Kasım 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, Türkiye, 5 - 08 Eylül 2018, ss.5-6
- IV. JÜVENİL DERMATOMİYOZİT HASTALARININ KAS BİYOPSİLERİNDE YARDIMCI T HÜCRE PROFİLLERİ  
SAĞ E., ÖZEN S., Kale G., TALİM B., TOPALOĞLU H.  
4. Cocuk Romatoloji Kongresi, Türkiye, 4 - 07 Nisan 2018
- V. Desmin Mutation with an ultra rare and unique phenotype: Genome editing for a patient specific zebrafish model  
Kayman Kürekçi G., Koyunlar C., Kural Mangit E., Talim B., Korkusuz P., Erdem Özdamar S., Puralı N., Dinçer P. R.  
keystone symposia Rare and Undiagnosed Diseases, Massachusetts, Amerika Birleşik Devletleri, 3 - 05 Ağustos 2017
- VI. Modeling of a unique desmin mutation in zebrafish by using genome editing brings new insights into desmin function  
KAYMAN KÜREKÇİ G., Koyunlar C., KURAL MANGIT E., TALİM B., ERGİN B., ÜNSAL İ., PURALI N., KORKUSUZ P., ERDEM ÖZDAMAR S., DİNÇER P. R.  
European Human Genetics Conference, 27 - 30 Mayıs 2017
- VII. The profile and natural history of congenital muscular dystrophies  
Ardicli D., Genc H. M., SEYHAN K., Kahraman A., Akcil M., TALİM B., Haliloglu G., YILMAZ O., Alemdaroglu İ., TOPALOĞLU H. A., et al.  
21st International Congress of the World-Muscle-Society, Granada, Nikaragua, 4 - 08 Ekim 2016, cilt.26
- VIII. Prenatal findings and autopsy examination in a newborn with multiple acyl CoA dehydrogenase deficiency  
DOKUZBOY S., TALİM B., YİĞİT Ş., SİVRİ H. S., TOKATLI A., DURSUN A., COŞKUN T.  
SSIEM ROMA, 6 - 09 Eylül 2016
- IX. Skeletal muscle expression of insulin like growth factor system elements in spinal muscular atrophy patients  
YEŞBEK KAYMAZ A., TALİM B., BORA G., YURTER H.  
5th International Congress of Myology, Lyon, Fransa, 14 - 18 Mart 2016
- X. Glutarik Asidemi Tip 2 Dismorfolojik İpuçları Veren Metabolik Bir Hastalık  
AKGÜN DOĞAN Ö., ÜNSAL Y., ŞİMŞEK KİPER P. Ö., ÜTİNE G. E., DURSUN A., TALİM B., YİĞİT Ş.  
3. Nörometabolik Dismorfoloji Sempozyumu, Türkiye, 10 - 12 Mart 2016
- XI. Spinal müsküler atrofi hastalarının iskelet kasında insülin benzeri büyümeye faktörleri ve bağlayıcı protein düzeylerinin araştırılması  
Yeşbek Kaymaz A., Bora G., Yurter H., Talim B.  
XIV. Ulusal Tibbi Biyoloji ve Genetik Kongresi, Muğla, Türkiye, 27 - 30 Ekim 2015
- XII. Çekirdek zarfı hastalıkları ile ilişkili yeni bir gen: TOR1AIP1 ve kas distrofisi  
Kayman Kürekçi G., Talim B., Puralı N., Dinçer P. R.  
14. Ulusal Tibbi Biyoloji ve Genetik Kongresi, Muğla, Türkiye, 27 Ekim 2015
- XIII. Inflammatory milieu of muscle biopsies and clinical features in juvenile dermatomyositis  
SAĞ E., ÖZEN S., KALE G., TOPALOĞLU H. A., TALİM B.  
20th International Congress of the World-Muscle-Society, Brighton, Birleşik Krallık, 30 Eylül - 04 Ekim 2015, cilt.25
- XIV. Etiological yield of muscle biopsy in the newborn period

- Serdaroglu E., Haliloglu G., TALİM B., YİĞİT Ş., YURDAKÖK M., Topaloglu H.  
20th International Congress of the World-Muscle-Society, Brighton, Birleşik Krallık, 30 Eylül - 04 Ekim 2015, cilt.25
- XV. **Spinal müsküler atrofi hastalarının iskeletkasında insulin benzeri büyümeye faktörleri ve bağlayıcı protein düzeylerinin araştırılması**  
YEŞBEK KAYMAZ A., TALİM B., BORA TATAR G., YURTER H.  
XIV. Ulusal Tibbi Biyoloji ve Genetik Kongresi, Fethiye, Türkiye, 27 Ekim 2015 - 30 Ekim 2014
- XVI. **Torsin A-interacting protein 1/Lamina-associated polypeptide 1B in a form of limb-girdle muscular dystrophy: a novel gene related to nuclear envelopathies**  
Kayman Kürekçi G., Hayta B., Talim B., Puralı N., Dinçer P. R.  
18. International Meeting of the World Muscle Society, California, Amerika Birleşik Devletleri, 05 Ekim 2013, ss.64
- XVII. **A novel desmin mutation causes autosomal recessive limb girdle muscular dystrophy without features of myofibrillar myopathy**  
Cetin N., Balci-Hayta B., Gundesli H., KORKUSUZ P., PURALI N., TALİM B., Tan E., Selcen D., Erdem-Ozdamar S., DİNÇER P. R.  
18th International Congress of the World-Muscle-Society (WMS), California, Amerika Birleşik Devletleri, 1 - 05 Ekim 2013, cilt.23, ss.851-852
- XVIII. **Formyl-methionyl-leucyl-phenylalanine (fMLP) peptide as an adjuvant in experimental autoimmune encephalomyelitis (EAE): modulation of neutrophil granulocytes**  
KILIÇ A., Tuncer-Kurne A., ESENDAĞLI G., Sayat G., TALİM B., KARABUDAK R.  
29th Congress of the European-Committee-for-Treatment-and-Research-in-Multiple-Sclerosis / 18th Annual Conference of Rehabilitation in MS, Copenhagen, Danimarka, 2 - 05 Ekim 2013, cilt.19, ss.136
- XIX. **The first prenatal diagnosis in Muscle-Eye-Brain Disease**  
Balci B., Celebi A., Talim B., Dincer P., Topaloglu H.  
11th International Congress on Neuromuscular Diseases, İstanbul, Türkiye, 2 - 07 Temmuz 2006, cilt.16
- XX. **Marinesco-Sjogren Syndrome and olivopontocerebellar hypoplasia with BAP/SIL mutations: report of a family with three siblings**  
Sonmez F. M., Gooding R., Talim B., Celep F., Kalaydjieva L., Topaloglu H.  
11th International Congress on Neuromuscular Diseases, İstanbul, Türkiye, 2 - 07 Temmuz 2006, cilt.16
- XXI. **Autosomal recessive limb-girdle muscular dystrophies (LGMD2s) in Turkey**  
Balci B., Talim B., Akcoeren Z., Caglar M., Kale G., Topaloglu H., Dincer P.  
11th International Congress on Neuromuscular Diseases, İstanbul, Türkiye, 2 - 07 Temmuz 2006, cilt.16
- XXII. **Prenatal diagnosis in laminin alpha-2 chain (merosin)-deficient congenital muscular dystrophy: a collective experience of 5 international centers**  
Vainzof M., Richard P., Herrmann R., Jimenez-Mallebrera C., Talim B., Yamamoto L., Ledeuil C., Mein R., Abbs S., Brockington M., et al.  
10th International Congress of the World-Muscle-Society, Iguassu Falls, Brezilya, 28 Eylül - 10 Ekim 2005, cilt.15, ss.704
- XXIII. **EUROSMART: motor function and muscle strength in 110 patients with spinal muscular atrophy**  
Merlini L., Dahna-Schwake C., Febrer A., Hausanova-Petruse I., Jedrzejonska M., Shapira Y., Taustein I., Luppi M., Tizzano E., Topaloglu H., et al.  
10th International Congress of the World-Muscle-Society, Iguassu Falls, Brezilya, 28 Eylül - 10 Ekim 2005, cilt.15, ss.724
- XXIV. **Focal segmental glomerulosclerosis associated with mitochondrial cytopathy: Report of two cases with special emphasis on podocytes**  
Gucer S., Talim B., Asan E., Ozen S., Kale G., Ozguc M., Caglar M.  
42nd Annual Meeting of the European-Renal-Association/European-Dialysis-and-Transplant-Association (ERA-EDTA), İstanbul, Türkiye, 4 - 07 Haziran 2005, cilt.20
- XXV. **A floppy infant with nemaline rods and plus**  
Talim B., Sabatelli P., Haliloglu G., Akcoren Z., Caglar M., Topaloglu H.  
9th International Congress of the World-Muscle-Society, Göteborg, İsveç, 1 - 04 Eylül 2004, cilt.14, ss.564-565
- XXVI. **Muscle pathology in childhood cases of calpainopathy**

- Talim B., Dincer P. R., Richard I., Aurino S., Akcoren Z., Haliloglu G., Kale G., Leturcq F., Nigro V., Topaloglu H.  
9th International Congress of the World-Muscle-Society, Göteborg, İsveç, 1 - 04 Eylül 2004, cilt.14, ss.605
- XXVII. **Apoptosis in Ullrich congenital muscular dystrophy**  
Talim B., Haliloglu G., Kale G., Topaloglu H.  
9th International Congress of the World-Muscle-Society, Göteborg, İsveç, 1 - 04 Eylül 2004, cilt.14, ss.615
- XXVIII. **Autosomal recessive limb-girdle muscular dystrophy with severe mental retardation: a new phenotype with glycosylation defects of alpha-dystroglycan**  
Dincer P. R., Balci B., Yuva Y., Talim B., Brockington M., Dincel D., Gerceker F., Haliloglu G., Atalay R., Yakicier C., et al.  
7th International Congress of the World-Muscle-Society, Rotterdam, Hollanda, 2 - 05 Ekim 2002, cilt.12, ss.721

## **Desteklenen Projeler**

TALİM B., Yükseköğretim Kurumları Destekli Proje, İnflamatuar Myofibroblastik Tümörlerde IgG4 ve Yardımcı T Hücreleri, 2014 - 2020

## **Metrikler**

Yayın: 148  
Atıf (WoS): 2546  
Atıf (Scopus): 2879  
H-İndeks (WoS): 25  
H-İndeks (Scopus): 26