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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. **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)
- II. **Early-onset juvenile dermatomyositis: A tertiary referral center experience and review of the literature**  
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Seminars in Arthritis and Rheumatism, vol.58, 2023 (SCI-Expanded)
- III. **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.  
Journal of neuromuscular diseases, vol.10, no.2, pp.279-291, 2023 (SCI-Expanded)
- IV. **Alterations in insulin-like growth factor system in spinal muscular atrophy**  
YEŞBEK KAYMAZ A., Bal S. K., Bora G., TALİM B., Ozon A., ALİKAŞIYOĞLU A., Topaloglu H., YURTER H.  
MUSCLE & NERVE, vol.66, no.5, pp.631-638, 2022 (SCI-Expanded)
- V. **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, vol.30, no.SUPPL 1, pp.140, 2022 (SCI-Expanded)

- VI. **A Rare Pediatric Case of Severe Rhabdomyolysis Owing to Dual Infection**  
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- VII. **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.  
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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. **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, vol.11, no.1, 2021 (SCI-Expanded)
- X. **Loss of C2orf69 defines a fatal autoinflammatory syndrome in humans and zebrafish that evokes a glycogen-storage-associated mitochondriopathy**  
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- XII. **Milk of calcium: A rare manifestation of juvenile dermatomyositis**  
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- XIII. **The Common miRNA Signatures Associated with Mitochondrial Dysfunction in Different Muscular Dystrophies**  
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- XIV. **Circulating Epstein-Barr virus DNA and cell-free DNA in pediatric lymphomas**  
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TURKISH JOURNAL OF PEDIATRICS, vol.62, no.4, pp.541-550, 2020 (SCI-Expanded)
- XV. **Determinants of Riboflavin Responsiveness in Multiple Acyl-CoA Dehydrogenase Deficiency**  
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- XVI. **Nemaline rod myopathy treated with L-tyrosine to relieve symptoms in a neonate**  
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- XVII. **Do not Miss Rare and Treatable Cause of Early-Onset Hemolytic Uremic Syndrome: Cobalamin C Deficiency**  
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- XVIII. **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.  
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- XIX. **LARGE expression in different types of muscular dystrophies other than dystroglycanopathy**  
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- XX. **Prenatal Diagnosis of Merosin-Deficient Muscular Dystrophy**  
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- XXI. **Non-immune hydrops fetalis: A retrospective analysis of 151 autopsies performed at a single center**  
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- XXIV. **Recessive PIEZO2 stop mutation causes distal arthrogryposis with distal muscle weakness, scoliosis and proprioception defects**  
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- XXV. **Congenital mirror movements in a patient with alpha-dystroglycanopathy due to a novel POMK mutation**  
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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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- XXVII. **Giant Omental Cyst (Lymphangioma) Mimicking Ascites and Tuberculosis**  
Karhan A. N., SOYER T., Gunes A., TALİM B., KARNAK İ., OĞUZ B., Temizel İ. N.  
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- XXVIII. **Riboflavin-Responsive and -Non-responsive Mutations in FAD Synthase Cause Multiple Acyl-CoA Dehydrogenase and Combined Respiratory-Chain Deficiency**  
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- XXIX. **Next generation sequencing in a large cohort of patients presenting with neuromuscular disease before or at birth**  
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- XXX. **Clinical characteristics of megaconial congenital muscular dystrophy due to choline kinase beta gene defects in a series of 15 patients**  
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- XXXI. **Promotion of experimental autoimmune encephalomyelitis upon neutrophil granulocytes' stimulation with formyl-methionyl-leucyl-phenylalanine (fMLP) peptide**  
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- XXXII. **SPEG Interacts with Myotubularin, and Its Deficiency Causes Centronuclear Myopathy with Dilated Cardiomyopathy**  
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- XXXIII. **Use of Whole-Exome Sequencing to Determine the Genetic Basis of Multiple Mitochondrial Respiratory Chain Complex Deficiencies**  
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- XXXIV. **Unusual splenic hemangioma of a pediatric patient: hypointense on T2-weighted image**  
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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. **Recessive TTN truncating mutations define novel forms of core myopathy with heart disease**  
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- XXXVII. **Vesiculopustular eruption in neonatal transient myeloproliferative disorder**  
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- XXXVIII. **Prenatal diagnosis and clinicopathologic examination of a case with diastematomyelia**  
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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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- XLII. **Serum alpha-fetoprotein levels in neonatal cholestasis**  
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- XLIII. **Multisystem fatal infantile disease caused by a novel homozygous EARS2 mutation**  
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- XLIV. **Solid tumors in Turkish children: a multicenter study**  
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- XLV. **Coexistence of two distinct intragenic dystrophin deletions in two maternal cousins with Duchenne Muscular Dystrophy**

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- XLVII. **Early corticosteroid treatment in 4 duchenne muscular dystrophy patients: 14-year follow-up**  
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- XLVIII. **Colchicine Protects against Hyperoxic Lung Injury in Neonatal Rats**  
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- L. **Nuclear factors involved in mitochondrial translation cause a subgroup of combined respiratory chain deficiency**  
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- LII. **Long-term follow-up in patients with congenital myasthenic syndrome due to CHAT mutations**  
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- LIII. **Double heterotopic pancreas and Meckel's diverticulum in a child: do they have a common origin?**  
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- LIV. **A RARE TYPE OF RENAL CELL CARCINOMA IN A GIRL: Hybrid Renal Cell Carcinoma**  
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- LV. **Sarcoid-like granulomas in common variable immunodeficiency**  
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- LVI. **An unusual presentation of Muscle-Eye-Brain disease: Severe eye abnormalities with mild muscle and brain involvement**  
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- LVII. **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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- LVIII. **Does Defective Apoptosis Play A Role in Cystic Fibrosis Lung Disease?**  
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- LXIII. **Pseudo-trisomy 13 in a fetus: further support for autosomal recessive inheritance**  
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- LXVI. **The myopathic form of coenzyme Q10 deficiency is caused by mutations in the electron-transferring-flavoprotein dehydrogenase (ETFDH) gene**  
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