Prof. Hacer DURMUŞ TEKÇE

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Education Information

Undergraduate, Istanbul University, Open And Dıstance Educatıon Faculty, Open Education Programs, Turkey 2016 - 2020
Expertise In Medicine, Istanbul University, Nöroloji, Turkey 2006 - 2010
Doctorate, Hacettepe University, Tıp Fakültesi, Turkey 2000 - 2006

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Dissertations

Expertise In Medicine, Okülofaringodistal Miyopatinin Klinik ve Genetik Özelliklerinin Belirlenmesi, Istanbul University, Nöroloji Anabilim Dalı, Nöromüsküler Hastalıklar Bilim Dalı, 2010

Research Areas

Medicine, Health Sciences, Internal Medicine Sciences, Neurology

Academic Titles / Tasks

Professor, Istanbul University, Istanbul Medical Faculty, Division of Medical Sciences , 2021 - Continues
Associate Professor, Istanbul University, Istanbul Medical Faculty, Division Of Medical Sciences , 2015 - 2021
Research Assistant, Newcastle College, 2009 - 2009

Jury Memberships

Appointment to Academic Staff - Associate Professorship, Appointment to Academic Staff - Associate Professorship, Istanbul University, May, 2023

Published journal articles indexed by SCI, SSCI, and AHCI

I. **Disease activity in chronic inflammatory demyelinating polyneuropathy: association between circulating B-cell subsets, cytokine levels, and clinical outcomes.**
Ozdag Acarli A. N., Tuzun E., Sanli E., Koral G., Akbayir E., Cakar A., Sirin N. G., Soysal A., Aysal F., Durmus H., et al.
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II. **Data from the European registry for patients with McArdle disease (EUROMAC): functional status and social participation**
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VI. **A novel homozygous loss-of-function variant in SOD1 causing progressive spastic tetraplegia and axial hypotonia**
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VIII. **Investigation of a Novel Recessive Mutation in Acetylcholine Receptor Epsilon-Subunit with Slow-Channel Kinetics Causing Congenital Myasthenia Reveals Subunit-Specific Contribution of Cys-Loop Length to Receptor Activation**
Shen X., Shen S., Zhao Y., Di L., Durmus H., Deymeer F., Bregman J., Selcen D., Engel A. G.
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IX. **Desmin Knock-Out Cardiomyopathy: A Heart on the Verge of Metabolic Crisis**
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X. **Clinical and genetic characteristics of Emery-Dreifuss muscular dystrophy patients from Turkey: 30 years longitudinal follow-up study.**
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XI. **Cerebellar ataxia, neuropathy and vestibular areflexia syndrome (canvas): an important cause of late-onset ataxia with unique clinical features**
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XII. **AN EXPLORATORY STUDY OF COGNITIVE INVOLVEMENT IN HEREDITARY ATTRV**
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XIII. **Phenotypical spectrum of SACS variants: Neuromuscular perspective of a complex**

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XIV. **Lumbar Spinal Stenosis: A Rare Presentation of Hereditary Transthyretin Amyloidosis**
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XX. **The Complex Genetic Landscape of Hereditary Ataxias in Turkey and Implications in Clinical Practice**
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XXI. **Diaphragmatic dysfunction at the first visit to a chest diseases outpatient clinic in 500 patients with amyotrophic lateral sclerosis**
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XXIII. **Late-onset TK2-Deficiency Patients from Turkey**
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XXIV. **Genotypic and phenotypic features of mutations in the HINT1 gene among Turkish patients with hereditary axonal neuropathy**
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Toepf A., Johnson K., Bates A., Phillips L., Chao K. R., England E. M., Laricchia K. M., Mullen T., Valkanas E., Xu L., et al.
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XXX. **Revisiting the complex architecture of ALS in Turkey: Expanding genotypes, shared phenotypes, molecular networks, and a public variant database**
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XXXI. **Four Individuals with a Homozygous Mutation in Exon 1f of thePLECGene and Associated Myasthenic Features**
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XXXIII. **Expanding the disease phenotype of ADSSL1-associated myopathy in non-Korean patients**
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XXXIV. **Late-onset generalized myasthenia gravis: clinical features, treatment, and outcome**
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XXXV. **A Val30Met sporadic familial amyloid polyneuropathy case with atypical presentation: upper limb onset of symptoms**
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XXXVI. **A novel homozygous FBXO38 variant causes an early-onset distal hereditary motor neuronopathy type IID.**
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XXXVII. **The first biallelic missense mutation in the FXN gene in a consanguineous Turkish family with Charcot-Marie-Tooth-like phenotype**
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XXXIX. **Mutation spectrum of 260 dystrophinopathy patients from Turkey and important highlights for genetic counseling**
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XL. **Familial Amyloid Polyneuropathy**
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XLI. **Extending the clinical and mutational spectrum of TRIM32-related myopathies in a non-Hutterite population**
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XLII. **Repetitive nerve stimulation and jitter measurement with disposable concentric needle electrode in newly diagnosed myasthenia gravis patients**
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XLV. **Congenital myasthenic syndromes in Turkey: Clinical clues and prognosis with long term follow-up**
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Scientific Refereeing

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Non Academic Experience

Başkale Devlet Hastanesi