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KiŐisel Bilgiler

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Eđitim Bilgileri

Lisans, İstanbul Üniversitesi, Açık Ve Uzaktan Eğitim Fakültesi, Açık Öğretim Programları, Türkiye 2016 - 2020

Tıpta Uzmanlık, İstanbul Üniversitesi, Nöroloji, Türkiye 2006 - 2010

Doktora, Hacettepe Üniversitesi, Tıp Fakültesi, Türkiye 2000 - 2006

Yabancı Diller

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Yaptığı Tezler

Tıpta Uzmanlık, Okülofaringodistal Miyopatinin Klinik ve Genetik Özelliklerinin Belirlenmesi, İstanbul Üniversitesi, Nöroloji Anabilim Dalı, Nöromüsküler Hastalıklar Bilim Dalı, 2010

AraŐtırma Alanları

Tıp, Sağlık Bilimleri, Dahili Tıp Bilimleri, Nöroloji

Akademik Unvanlar / Görevler

Prof. Dr., İstanbul Üniversitesi, İstanbul Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2021 - Devam Ediyor

Doç. Dr., İstanbul Üniversitesi, İstanbul Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2015 - 2021

AraŐtırma Görevlisi, Newcastle College, 2009 - 2009

Jüri Üyelikleri

Akademik Kadroya Atama-Doçentlik, Akademik Kadroya Atama-Doçentlik, İstanbul Üniversitesi, Mayıs, 2023

SCI, SSCI ve AHCI İndekslerine Giren Dergilerde Yayınlanan Makaleler

- Thymic gene expression analysis reveals a potential link between HIF-1A and Th17/Treg imbalance

in thymoma associated myasthenia gravis

Altınönder İ., Kaya M., Yentür S. P., Çakar A., Durmuş H., Yegen G., Özkan B., Parman Y., Sawalha A. H., Saruhan-Direskeneli G.

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- II. **In vitro modulation of T cells in myasthenia gravis by low-dose IL-2**
Çebi M., Çakar A., Durmuş H., Akan O., Aysal F., Parman Y., Saruhan-Direskeneli G.
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- III. **Disease activity in chronic inflammatory demyelinating polyneuropathy: association between circulating B-cell subsets, cytokine levels, and clinical outcomes**
Ozdogan 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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- IV. **Data from the European registry for patients with McArdle disease (EUROMAC): functional status and social participation**
Karazi W., Scalco R. S., Stemmerik M. G., Løkken N., Lucia A., Santalla A., Martinuzzi A., Vavla M., Reni G., Toscano A., et al.
Orphanet Journal of Rare Diseases, cilt.18, sa.1, 2023 (SCI-Expanded)
- V. **Phenotypic features of RETREG1-related hereditary sensory autonomic neuropathy**
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- VI. **Thymoma patients with or without myasthenia gravis have increased Th17 cells, IL-17 production and ICOS expression**
Cebi M., Cakar A., Erdogdu E., Durmus-Tekce H., Yegen G., Ozkan B., Parman Y., Saruhan-Direskeneli G.
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- VII. **Expert opinion on the diagnostic odyssey and management of late-onset Pompe disease: a neurologist's perspective**
ERDEM ÖZDAMAR S., KOÇ A. F., Durmus Tekce H., Kotan D., Ekmekci A. H., ŞENGÜN İ. Ş., Yuceyar A. N., Uluc K.
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- VIII. **A novel homozygous loss-of-function variant in SOD1 causing progressive spastic tetraplegia and axial hypotonia**
Çakar A., Pekbilir E., CEYLANER S., Durmuş H., Battaloğlu E., Şahin U., Parman Y.
Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, cilt.24, sa.5-6, ss.535-538, 2023 (SCI-Expanded)
- IX. **Disease activity in chronic inflammatory demyelinating polyneuropathy: A comparative study of clinical and skin biopsy markers**
Acarli A. N. O., Unverengil G., Sirin N. G., Cakar A., Durmus H., Parman Y.
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- X. **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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- XI. **Desmin Knock-Out Cardiomyopathy: A Heart on the Verge of Metabolic Crisis**
Elsnicova B., Hornikova D., Tibenska V., Kolar D., Tlapakova T., Schmid B., Mallek M., Eggers B., Schloetzer-Schrehardt U., Peeva V., et al.
INTERNATIONAL JOURNAL OF MOLECULAR SCIENCES, cilt.23, sa.19, 2022 (SCI-Expanded)
- XII. **Clinical and genetic characteristics of Emery-Dreifuss muscular dystrophy patients from Turkey: 30 years longitudinal follow-up study.**
Yunisova G., CEYLANER S., Oflazer P., Deymeer F., Parman Y. G., Durmus H.
Neuromuscular disorders : NMD, cilt.32, sa.9, ss.718-727, 2022 (SCI-Expanded)
- XIII. **Cerebellar ataxia, neuropathy and vestibular areflexia syndrome (canvas): an important cause of late-onset ataxia with unique clinical features**
ÇAKAR A., Sahin E., Tezel S., Candayan A., Samanci B., BATTALOĞLU E., Basak A. N., Bilgic B., Hanagasi H. A., Durmus

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- XIV. **AN EXPLORATORY STUDY OF COGNITIVE INVOLVEMENT IN HEREDITARY ATTRV**
Durmus H., Cakar A., Demirci H., Parman Y.
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- XV. **Phenotypical spectrum of SACS variants: Neuromuscular perspective of a complex neurodegenerative disorder**
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- XVI. **Lumbar Spinal Stenosis: A Rare Presentation of Hereditary Transthyretin Amyloidosis**
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- XVII. **Rare slow channel congenital myasthenic syndromes without repetitive compound muscle action potential and dramatic response to low dose fluoxetine**
Durmus H., Sticht H., Ceylaner S., Hashemolhosseini S., Deymeer F.
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- XVIII. **An Exploratory Study of Cognitive Involvement in Hereditary Transthyretin Amyloidosis**
Durmus H., Cakar A., Demirci H., ALAYLIOĞLU M., GEZEN AK D., DURSUN E., Gulsen Parman Y.
ACTA NEUROLOGICA SCANDINAVICA, cilt.144, sa.6, ss.640-646, 2021 (SCI-Expanded)
- XIX. **Genetic Survey of Autosomal Recessive Peripheral Neuropathy Cases Unravels High Genetic Heterogeneity in a Turkish Cohort**
Candayan A., Çakar A., Yunisova G., Özdağ Acarlı A. N., Atkinson D., Topaloğlu P., Durmuş H., Yapıcı Z., Jordanova A., Parman Y., et al.
NEUROLOGY-GENETICS, cilt.7, sa.5, 2021 (SCI-Expanded)
- XX. **Cerebellar ataxia, neuropathy and vestibular areflexia syndrome (Canvas) is an important cause of late-onset ataxia**
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- XXI. **Episodic psychosis, ataxia, motor neuropathy with pyramidal signs (PAMP syndrome) caused by a novel mutation in ADPRHL2 (AHR3)**
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- XXII. **The Complex Genetic Landscape of Hereditary Ataxias in Turkey and Implications in Clinical Practice**
Vural A., Sinsir G., Tekgul S., Kocoglu C., Akcimen F., Kartal E., Sen N. E., Lahut S., Omur O., Saner N., et al.
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- XXIII. **Diaphragmatic dysfunction at the first visit to a chest diseases outpatient clinic in 500 patients with amyotrophic lateral sclerosis**
Pihtili A., Bingol Z., Durmus H., Parman Y., Kiyan E.
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- XXIV. **SOD1 Mutation: A Single Center Experience**
ÇAKAR A., DURMUŞ TEKÇE H., PARMAN F. Y.
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- XXV. **Late-onset TK2-Deficiency Patients from Turkey**
DURMUŞ TEKÇE H., ÇAKAR A., PARMAN F. Y.
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- XXVI. **Genotypic and phenotypic features of mutations in the HINT1 gene among Turkish patients with hereditary axonal neuropathy**
Acarlı A. O., Cakar A., Candayan A., Durmus H., Ceylaner S., Matur Z., Oge A., Parman Y.
JOURNAL OF THE PERIPHERAL NERVOUS SYSTEM, cilt.26, sa.1, ss.124-125, 2021 (SCI-Expanded)
- XXVII. **The treatment effect on peripheral B cell markers in antibody positive myasthenia gravis patients**

- YILMAZ V., TÜZÜN E., DURMUŞ TEKÇE H., Oflazer P., Aysal F., Parman Y., Gungor-Tuncer O., Deymeer F., Saruhan-Direskeneli G.
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- XXVIII. **Cognition of the mothers of patients with Duchenne muscular dystrophy.**
Demirci H., Durmus H., Toksoy G., Uslu A., Parman Y., Hanagasi H. A.
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- XXIX. **Data from the European registry for patients with McArdle disease and other muscle glycogenoses (EUROMAC)**
Scalco R. S., Lucia A., Santalla A., Martinuzzi A., Vavla M., Reni G., Toscano A., Musumeci O., Voermans N. C., Kouwenberg C. V., et al.
ORPHANET JOURNAL OF RARE DISEASES, cilt.15, sa.1, 2020 (SCI-Expanded)
- XXX. **Correlations between radiographic spinopelvic parameters and health-related quality of life: A prospective evaluation of 37 patients with facioscapulohumeral muscular dystrophy**
Bayram S., Kendirci A. Ş., Karalar Ş., Durmuş Tekçe H., Parman F. Y., Akgül T., Durmaz H.
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- XXXI. **Sequential targeted exome sequencing of 1001 patients affected by unexplained limb-girdle weakness**
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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- XXXII. **Revisiting the complex architecture of ALS in Turkey: Expanding genotypes, shared phenotypes, molecular networks, and a public variant database**
Tunca C., Seker T., Akcimen F., Coskun C., Bayraktar E., Palvadeau R., Zor S., Kocoglu C., Kartal E., Sen N. E., et al.
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- XXXIII. **Four Individuals with a Homozygous Mutation in Exon 1f of the PLECGene and Associated Myasthenic Features**
Mroczek M., DURMUŞ TEKÇE H., Topf A., Parman Y., Straub V.
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- XXXIV. **CD4+ T Cells of Myasthenia Gravis Patients Are Characterized by Increased IL-21, IL-4, and IL-17A Productions and Higher Presence of PD-1 and ICOS**
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- XXXV. **Expanding the disease phenotype of ADSSL1-associated myopathy in non-Korean patients**
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- XXXVI. **Late-onset generalized myasthenia gravis: clinical features, treatment, and outcome**
Yildiz Celik S., DURMUŞ TEKÇE H., YILMAZ V., SARUHAN DİRESKENELİ G., Gulsen Parman Y., Serdaroglu Oflazer P., Deymeer F.
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- XXXVII. **A Val30Met sporadic familial amyloid polyneuropathy case with atypical presentation: upper limb onset of symptoms**
Sahin E., Cakar A., Durmus-Tekce H., Parman Y.
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- XXXVIII. **A novel homozygous FBX038 variant causes an early-onset distal hereditary motor neuronopathy type IID.**
Akcimen F., Vural A., Durmus H., Cakar A., Houlden H., Parman Y. G., Basak A. N.
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- XXXIX. **The first biallelic missense mutation in the FXN gene in a consanguineous Turkish family with Charcot-Marie-Tooth-like phenotype**
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- XL. **Relation of HLA-DRB1 to IgG4 autoantibody and cytokine production in muscle-specific tyrosine kinase myasthenia gravis (MuSK-MG).**
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- XLII. **Mutation spectrum of 260 dystrophinopathy patients from Turkey and important highlights for genetic counseling**
Toksoy G., Durmus H., Aghayev A., Bagirova G., Rustemoglu B. S., Basaran S., Avci S., Karaman B., Parman Y., Altunoglu U., et al.
NEUROMUSCULAR DISORDERS, sa.8, ss.601-613, 2019 (SCI-Expanded)
- XLIII. **Extending the clinical and mutational spectrum of TRIM32-related myopathies in a non-Hutterite population**
Johnson K., De Ridder W., Topf A., Bertoli M., Phillips L., De Jonghe P., Baets J., Deconinck T., Stojanovic V. R., Peric S., et al.
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- XLIV. **Repetitive nerve stimulation and jitter measurement with disposable concentric needle electrode in newly diagnosed myasthenia gravis patients**
Sirin N. G., Orhan E., Durmus H., Deymeer F., Baslo M. B.
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- XLV. **Increased costimulatory molecule expression of thymic and peripheral B cells and a sensitivity to IL-21 in myasthenia gravis**
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- XLVI. **Detection of variants in dystroglycanopathy-associated genes through the application of targeted whole-exome sequencing analysis to a large cohort of patients with unexplained limb-girdle muscle weakness**
Johnson K., Bertoli M., Phillips L., Topf A., Van den Bergh P., Vissing J., Witting N., Nafissi S., Jamal-Omidi S., Lusakowska A., et al.
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- XLVII. **Congenital myasthenic syndromes in Turkey: Clinical clues and prognosis with long term follow-up**
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- XLVIII. **A database for screening and registering late onset Pompe disease in Turkey**
GOKYIGIT M. C., Ekmekci H., Durmus H., Karll N., Koseoglu E., AYSAL F., Kotan D., ALI A., Koytak P. K., Karasoy H., et al.
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- XLIX. **Mutations causing congenital myasthenia reveal principal coupling pathway in the acetylcholine receptor epsilon-subunit**
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- L. **Myophosphorylase (PYGM) mutations determined by next generation sequencing in a cohort from Turkey with McArdle disease**
Inal-Gultekin G., Toptas-Hekimoglu B., Gormez Z., Gelisin O., Durmus H., ERGUNER B., DEMİRCİ H., SAGIROGLU M. S., Parman Y., Deymeer F., et al.
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- LI. **Elevated IL-4 and IFN- γ Levels in Muscle Tissue of Patients with Dermatomyositis**
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- LII. **MUSCLE MAGNETIC RESONANCE IMAGING IN SPINAL MUSCULAR ATROPHY TYPE 3: SELECTIVE AND PROGRESSIVE INVOLVEMENT**
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- LIII. **Jitter measurement using a concentric needle in 133 patients with myasthenia gravis: A retrospective analysis**
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- LIV. **Neuromuscular endplate pathology in recessive desminopathies: Lessons from man and mice**
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- LV. **Genotypic and phenotypic presentation of transthyretin-related familial amyloid polyneuropathy (TTR-FAP) in Turkey.**
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- LVI. **Genetic heterogeneity within the HLA region in three distinct clinical subgroups of myasthenia gravis.**
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- LVII. **Titin antibodies in "seronegative" myasthenia gravis--A new role for an old antigen.**
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- LVIII. **Vocal Cord Paralysis and Hypercapnic Respiratory Failure in a Patient with Familial Amyloidotic Polyneuropathy**
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- LIX. **Reduced muscle mitochondrial enzyme activity in MuSK-immunized mice.**
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- LX. **MuSK autoantibodies in myasthenia gravis detected by cell based assay--A multinational study.**
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- LXI. **Differential cytokine changes in patients with myasthenia gravis with antibodies against AChR and MuSK**
Yilmaz V., Oflazer P., AYSAL F., Durmus H., Poulas K., Yentur S. P., Gulsen-Parman Y., TZARTOS S., MARX A., Tuzun E., et al.
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- LXII. **Regulatory function of CD4+CD25++ T cells in patients with myasthenia gravis is associated with phenotypic changes and STAT5 signaling: 1,25-Dihydroxyvitamin D3 modulates the suppressor activity.**
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- LXIII. **The distinct genetic pattern of ALS in Turkey and novel mutations**
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- LXIV. **Prevalence of Parkinson's disease in Baskale, Turkey: a population based study**
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- LXV. **Regulatory function of CD4+CD25++ T cells in patients with myasthenia gravis is associated with phenotypic changes and STAT5 signaling:1,25-Dihydroxyvitamin D3 modulates the suppressor activity**
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- LXVI. **Association between restless leg syndrom and slow coronary flow**
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- LXVII. **A comprehensive analysis of the epidemiology and clinical characteristics of anti-LRP4 in myasthenia gravis**
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- LXVIII. **Prevalence of Parkinson's disease in Baskale, Turkey: A population-based study**
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- LXIX. **Comparative clinical characteristics of early- and adult-onset multiple sclerosis patients with seizures**
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