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KiŐisel Bilgiler

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Uluslararası AraŐtırmacı ID'leri

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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. **Current insights in ultra-rare adenylosuccinate synthetase 1 myopathy – meeting report on the First Clinical and Scientific Conference. 3 June 2024, National Centre for Advancing Translational Science, Rockville, Maryland, the United States of America**
Rybalka E., Park H. J., Nalini A., Baskar D., Polavarapu K., Durmus H., Xia Y., Wan L., Shieh P. B., Moghadaszadeh B., et al.
Orphanet Journal of Rare Diseases, cilt.19, sa.1, 2024 (SCI-Expanded)
- III. **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.
European Journal of Immunology, cilt.54, sa.11, 2024 (SCI-Expanded)
- IV. **Detection of gene variants associated with recessive limb-girdle muscular weakness and Pompe disease in a global cohort of patients through the application of next-generation sequencing analysis**
Bevilacqua J. A., Al-Salti A. M., Al Madani A., Alves da Fonseca A., Durmus H., Chai J., Alshehri A., Ribeiro M. G., Sgobbi P., Nikitin S. S., et al.
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- V. **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.
CLINICAL AND EXPERIMENTAL IMMUNOLOGY, cilt.215, sa.1, ss.65-78, 2024 (SCI-Expanded)
- VI. **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.
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- VII. **Phenotypic features of RETREG1-related hereditary sensory autonomic neuropathy**
Çakar A., Bağirova G., Durmuş H., Uyguner O., Parman Y.
Journal of the Peripheral Nervous System, cilt.28, sa.3, ss.351-358, 2023 (SCI-Expanded)
- VIII. **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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- IX. **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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- X. **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)
- XI. **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.
MUSCLE & NERVE, cilt.66, sa.6, ss.736-743, 2022 (SCI-Expanded)
- XII. **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)
- XIII. **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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- XIV. **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)
- XV. **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 H., et al.
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- XVI. **AN EXPLORATORY STUDY OF COGNITIVE INVOLVEMENT IN HEREDITARY ATTRV**
Durmus H., Cakar A., Demirci H., Parman Y.
JOURNAL OF THE PERIPHERAL NERVOUS SYSTEM, cilt.27, 2022 (SCI-Expanded)
- XVII. **Phenotypical spectrum of SACS variants: Neuromuscular perspective of a complex neurodegenerative disorder**
Cakar A., Inci M., Acarli A. N. O., Comu S., Candayan A., BATTALOĞLU E., Tekgul S., Basak A. N., Durmus H., Parman Y.
ACTA NEUROLOGICA SCANDINAVICA, cilt.145, ss.619-626, 2022 (SCI-Expanded)
- XVIII. **Lumbar Spinal Stenosis: A Rare Presentation of Hereditary Transthyretin Amyloidosis**
Cakar A., Atmaca M. M., Kotan D., Durmus H., Deymeer F., Oflazer P., Parman Y.
NOROPSIKIYATRI ARSIVI-ARCHIVES OF NEUROPSYCHIATRY, cilt.59, sa.1, ss.77-79, 2022 (SCI-Expanded)
- XIX. **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.
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- XX. **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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- XXI. **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.
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- XXII. **Episodic psychosis, ataxia, motor neuropathy with pyramidal signs (PAMP syndrome) caused by a novel mutation in ADPRHL2 (AHR3)**
Durmus H., Mertoglu E., Sticht H., Ceylaner S., Kulaksizoglu I. B., Hashemolhosseini S., Ucar E. O., Parman Y.
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- XXIII. **Cerebellar ataxia, neuropathy and vestibular areflexia syndrome (Canvas) is an important cause of late-onset ataxia**
Cakar A., Sahin E., Tezel S., Candayan A., Samanci B., BATTALOĞLU E., Basak A. N., Bilgic B., Hanagasi H., Durmus H., et al.
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- XXIV. **The Complex Genetic Landscape of Hereditary Ataxias in Turkey and Implications in Clinical Practice**
Vural A., Simsir G., Tekgul S., Kocoglu C., Akcimen F., Kartal E., Sen N. E., Lahut S., Omur O., Saner N., et al.
MOVEMENT DISORDERS, cilt.36, ss.1676-1688, 2021 (SCI-Expanded)
- XXV. **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., Kiyani E.
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- XXVI. **Late-onset TK2-Deficiency Patients from Turkey**
DURMUŞ TEKÇE H., ÇAKAR A., PARMAN F. Y.
NEUROLOGY, sa.15, 2021 (SCI-Expanded)
- XXVII. **SOD1 Mutation: A Single Center Experience**
ÇAKAR A., DURMUŞ TEKÇE H., PARMAN F. Y.
NEUROLOGY, sa.15, 2021 (SCI-Expanded)
- XXVIII. **Genotypic and phenotypic features of mutations in the HINT1 gene among Turkish patients with hereditary axonal neuropathy**
Acarli 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)
- XXIX. **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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- XXX. **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)
- XXXI. **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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- XXXII. **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.
Clinical Neurology and Neurosurgery, cilt.198, 2020 (SCI-Expanded)
- XXXIII. **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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- XXXIV. **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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- XXXV. **Four Individuals with a Homozygous Mutation in Exon 1f of the PLECGene and Associated Myasthenic Features**
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- XXXVI. **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**
Cebi M., DURMUŞ TEKÇE H., Aysal F., ÖZKAN B., Gul G. E., ÇAKAR A., Hocaoglu M., Mercan M., YENTÜR S. P., Tutuncu M., et al.
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- XXXVII. **Expanding the disease phenotype of ADSSL1-associated myopathy in non-Korean patients**
Mroczek M., DURMUŞ TEKÇE H., Bijarnia-Mahay S., Topf A., Ghaoui R., Bryen S., Duff J., England E., Cooper S. T., MacArthur D. G., et al.
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- XXXVIII. **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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- XXXIX. **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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- XL. **A novel homozygous FBX038 variant causes an early-onset distal hereditary motor neuropathy type IID.**
Akcimen F., Vural A., Durmus H., Cakar A., Houlden H., Parman Y. G., Basak A. N.
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- XLI. **The first biallelic missense mutation in the FXN gene in a consanguineous Turkish family with Charcot-Marie-Tooth-like phenotype**
Candayan A., Yunisova G., Cakar A., Durmus H., Basak A. N., Parman Y., BATTALOĞLU E.
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- XLII. **Relation of HLA-DRB1 to IgG4 autoantibody and cytokine production in muscle-specific tyrosine kinase myasthenia gravis (MuSK-MG).**
ÇEBİ M., DURMUŞ H., YILMAZ V., YENTÜR S. P., AYSAL F., OFLAZER P., PARMAN Y., DEYMEER F., Saruhan-Direskeneli G.
Clinical and experimental immunology, cilt.197, ss.214-221, 2019 (SCI-Expanded)
- XLIII. **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)
- XLIV. **Familial Amyloid Polyneuropathy**
Cakar A., Durmus-Tekce H., Parman Y.
NOROSIKIYATRI ARSIVI-ARCHIVES OF NEUROPSYCHIATRY, cilt.56, sa.2, ss.150-156, 2019 (SCI-Expanded)
- XLV. **Extending the clinical and mutational spectrum of TRIM32-related myopathies in a non-Hutterite population**
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- XLVI. **Increased costimulatory molecule expression of thymic and peripheral B cells and a sensitivity to IL-21 in myasthenia gravis**
Hocaoglu M., Durmus H., Ozkan B., Yentur S. P., Dogan O., Parman Y., Deymeer F., Saruhan-Direskeneli G.
JOURNAL OF NEUROIMMUNOLOGY, cilt.323, ss.36-42, 2018 (SCI-Expanded)
- XLVII. **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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- XLVIII. **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.
SKELETAL MUSCLE, cilt.8, 2018 (SCI-Expanded)
- XLIX. **Congenital myasthenic syndromes in Turkey: Clinical clues and prognosis with long term follow-up**
Durmus H., SHEN X., Serdaroglu-Oflazer P., Kara B., Parman-Gulsen Y., Ozdemir C., BRENGMAN J., Deymeer F., ENGEL A. G.
NEUROMUSCULAR DISORDERS, cilt.28, sa.4, ss.315-322, 2018 (SCI-Expanded)
- L. **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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- LI. **Mutations causing congenital myasthenia reveal principal coupling pathway in the acetylcholine receptor epsilon-subunit**
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- LII. **Myophosphorylase (PYGM) mutations determined by next generation sequencing in a cohort from Turkey with McArdle disease**
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- LIII. **Elevated IL-4 and IFN- γ Levels in Muscle Tissue of Patients with Dermatomyositis**
Giris M., Durmus H., Yetimler B., Tasli H., Parman Y., Tuzun E.
IN VIVO, sa.4, ss.657-660, 2017 (SCI-Expanded)
- LIV. **MUSCLE MAGNETIC RESONANCE IMAGING IN SPINAL MUSCULAR ATROPHY TYPE 3: SELECTIVE AND PROGRESSIVE INVOLVEMENT**
Durmus H., Yilmaz R. S., Gulsen-Parman Y., Oflazer-Serdaroglu P., CUTTINI M., Dursun M., Deymeer F.
MUSCLE & NERVE, cilt.55, sa.5, ss.651-656, 2017 (SCI-Expanded)
- LV. **Jitter measurement using a concentric needle in 133 patients with myasthenia gravis: A retrospective analysis**
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Journal of Neurological Sciences, cilt.34, sa.3, ss.207-214, 2017 (SCI-Expanded)
- LVI. **Neuromuscular endplate pathology in recessive desminopathies: Lessons from man and mice**
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- LVII. **Genotypic and phenotypic presentation of transthyretin-related familial amyloid polyneuropathy (TTR-FAP) in Turkey.**
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- LVIII. **Genetic heterogeneity within the HLA region in three distinct clinical subgroups of myasthenia gravis.**
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- LIX. **Titin antibodies in "seronegative" myasthenia gravis--A new role for an old antigen.**
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- LX. **Vocal Cord Paralysis and Hypercapnic Respiratory Failure in a Patient with Familial Amyloidotic Polyneuropathy**
PIHTILI A., Bingol Z., Durmus H., Parman Y., Kiyan E.
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- LXI. **Reduced muscle mitochondrial enzyme activity in MuSK-immunized mice.**
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- LXII. **MuSK autoantibodies in myasthenia gravis detected by cell based assay--A multinational study.**
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- LXIII. 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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- LXIV. 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.**
Alahgholi-Hajibehzad M., Oflazer P., Aysal F., Durmus H., Gulsen-Parman Y., MARX A., Deymeer F., Saruhan-Direskeneli G.
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- LXV. The distinct genetic pattern of ALS in Turkey and novel mutations**
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- LXVI. Prevalence of Parkinson's disease in Baskale, Turkey: a population based study**
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- LXVII. 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**
Alahgholi-Hajibehzad M., OFLAZER Z. P., Aysal F., PARMAN F. Y., DURMUŞ TEKÇE H., Marx A., DEYMEER F., SARUHAN DİRESKENELİ G.
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