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Education Information

Expertise In Medicine, Université Paris-Sud: Paris XI, Tıp Fakültesi , Nöroloji, France 1988 - 1994

Doctorate, Istanbul University, Istanbul Medical Faculty, Turkey 1978 - 1984

Foreign Languages

French, C1 Advanced

English, C1 Advanced

Dissertations

Expertise In Medicine, Familial Amiloyid Nöropatinin Klinikopatolojik İncelenmesi, Université Paris-Sud: Paris XI, Dahili Tıp Bilimleri/Nöroloji, Nöroloji, 1992

Research Areas

Medicine, Health Sciences, Internal Medicine Sciences, Neurology

Academic Titles / Tasks

Professor, Istanbul University, Istanbul Medical Faculty, Division of Medical Sciences , 1995 - Continues

Published journal articles indexed by SCI, SSCI, and AHCI

- Thymic gene expression analysis reveals a potential link between HIF-1A and Th17/Treg imbalance in thymoma associated myasthenia gravis**
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- II. **Disease activity in chronic inflammatory demyelinating polyneuropathy: association between circulating B-cell subsets, cytokine levels, and clinical outcomes.**
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- III. **Genetic landscape of congenital insensitivity to pain and hereditary sensory and autonomic neuropathies.**
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- IV. **Eplontersen for Hereditary Transthyretin Amyloidosis With Polyneuropathy**
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- 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**
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- VII. **Characteristics of Patients with Hereditary Transthyretin Amyloidosis-Polyneuropathy (ATTRv-PN) in NEURO-TTTransform, an Open-label Phase 3 Study of Eplontersen**
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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, vol.24, no.5-6, pp.535-538, 2023 (SCI-Expanded)
- IX. **A multicentric study of the disease risks and first manifestations in Hereditary transthyretin amyloidosis (ATTRv): insights for an earlier diagnosis**
Planté-Bordeneuve V., Gorram F., Olsson M., Anan I., Mazzeo A., Gentile L., Cisneros-Barroso E., Gonzalez-Moreno J., Losada I., Waddington-Cruz M., et al.
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- X. **Clinical and genetic profile of patients enrolled in the Transthyretin Amyloidosis Outcomes Survey (THAOS): 14-year update**
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- XI. **Disease activity in chronic inflammatory demyelinating polyneuropathy: A comparative study of clinical and skin biopsy markers**
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- XII. **Clinical and genetic characteristics of Emery-Dreifuss muscular dystrophy patients from Turkey: 30 years longitudinal follow-up study.**
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- 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. **NOVEL VARIANTS BROADEN THE MUTATIONAL SPECTRUM OF HEREDITARY SENSORY AND AUTONOMIC NEUROPATHY DISORDERS**
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- XV. **AN EXPLORATORY STUDY OF COGNITIVE INVOLVEMENT IN HEREDITARY ATTRV**
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- XVI. **CHARACTERISTICS OF PATIENTS WITH HEREDITARY TRANSTHYRETIN AMYLOIDOSIS-POLYNEUROPATHY (ATTRV PN) IN NEURO-TTRANSFORM, A PHASE 3 STUDY OF EPLONTERSEN**
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- XVIII. **Phenotypical spectrum of SACS variants: Neuromuscular perspective of a complex neurodegenerative disorder**
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- XIX. **Genetics of Pain: Novel variants identified by the European Network on Inherited Sensory Neuropathies and Insensitivity to Pain (ENISNIP)**
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- XX. **Screening of SORD mutations in a CMT cohort expands the clinical spectrum of SORD-related neuropathy**
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- XXII. **Bi-allelic variants in neuronal cell adhesion molecule cause a neurodevelopmental disorder characterized by developmental delay, hypotonia, neuropathy/spasticity**
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- XXIII. **Clinical and Genetic Survey for Charcot-Marie-Tooth Neuropathy Based on the Findings in Turkey, a Country with a High Rate of Consanguineous Marriages**
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- XXIV. **An Exploratory Study of Cognitive Involvement in Hereditary Transthyretin Amyloidosis**
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- XXV. **Genetic Survey of Autosomal Recessive Peripheral Neuropathy Cases Unravels High Genetic Heterogeneity in a Turkish Cohort**
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- XXVI. **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.
NEUROLOGICAL SCIENCES, vol.42, no.9, pp.3871-3878, 2021 (SCI-Expanded)
- XXVII. **Cerebellar ataxia, neuropathy and vestibular areflexia syndrome (Canvas) is an important cause of late-onset ataxia**
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- XXVIII. **Screening of SORD mutations in a CMT cohort expands the clinical spectrum of SORD-related neuropathy**
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- XXIX. **The Complex Genetic Landscape of Hereditary Ataxias in Turkey and Implications in Clinical Practice**
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- XXXI. **SOD1 Mutation: A Single Center Experience**
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- XXXII. **Late-onset TK2-Deficiency Patients from Turkey**
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- XXXIII. **Dysregulation of myelin synthesis and actomyosin function underlies aberrant myelin in CMT4B1 neuropathy**
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- XXXIV. **Genotypic and phenotypic features of mutations in the HINT1 gene among Turkish patients with hereditary axonal neuropathy**
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- XXXV. **The treatment effect on peripheral B cell markers in antibody positive myasthenia gravis patients**
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- XXXVI. **Correlations between radiographic spinopelvic parameters and health-related quality of life: A prospective evaluation of 37 patients with facioscapulohumeral muscular dystrophy**
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- XXXVII. **Revisiting the complex architecture of ALS in Turkey: Expanding genotypes, shared phenotypes, molecular networks, and a public variant database**
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- XXXVIII. **Clinical and genetic aspects of hereditary spastic paraplegia in patients from Turkey**

- Akcakaya N. H., Ak B. O., Gonzalez M. A., Zuchner S., BATTALOĞLU E., Parman Y.
NEUROLOGIA I NEUROCHIRURGIA POLSKA, vol.54, no.2, pp.176-184, 2020 (SCI-Expanded)
- XXXIX. **A Val30Met sporadic familial amyloid polyneuropathy case with atypical presentation: upper limb onset of symptoms**
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- XL. **A novel homozygous FBXO38 variant causes an early-onset distal hereditary motor neuropathy type IID.**
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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**
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- XLII. **Linkage analysis and whole exome sequencing reveals AHNAK2 as a novel genetic cause for autosomal recessive CMT in a Malaysian family**
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- XLIII. **Mutation spectrum of 260 dystrophinopathy patients from Turkey and important highlights for genetic counseling**
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- XLIV. **Assessment of patients with hereditary transthyretin amyloidosis - understanding the impact of management and disease progression**
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- XLV. **A multicenter retrospective study of charcot-marie-tooth disease type 4B (CMT4B) associated with mutations in myotubularin-related proteins (MTMRs)**
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- XLVIII. **Turkish version of the Motor Function Measure Scale (MFM-32) for neuromuscular diseases: a cross-cultural adaptation, reliability, and validity study**
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- XLIX. **Myophosphorylase (PYGM) mutations determined by next generation sequencing in a cohort from Turkey with McArdle disease**
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- LII. **Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis**
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- LIII. **Neuromuscular endplate pathology in recessive desminopathies: Lessons from man and mice**
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- LIV. **Genotypic and phenotypic presentation of transthyretin-related familial amyloid polyneuropathy (TTR-FAP) in Turkey.**
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- LV. **Volumetric differences suggest involvement of cerebellum and brainstem in chronic migraine**
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- LVI. **Optimizing the management of transthyretin familial amyloid polyneuropathy in Europe: early diagnosis and effective care.**
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- LVII. **Sixty years of transthyretin familial amyloid polyneuropathy (TTR-FAP) in Europe: where are we now? A European network approach to defining the epidemiology and management patterns for TTR-FAP**
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- LVIII. **Vocal Cord Paralysis and Hypercapnic Respiratory Failure in a Patient with Familial Amyloidotic Polyneuropathy**
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- LIX. **Exome Sequence Analysis Suggests that Genetic Burden Contributes to Phenotypic Variability and Complex Neuropathy**
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- LX. **Transcriptional regulator PRDM12 is essential for human pain perception**
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- LXIII. **The distinct genetic pattern of ALS in Turkey and novel mutations**
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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**
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- LXV. **Unraveling the genetic landscape of autosomal recessive Charcot-Marie-Tooth neuropathies using a homozygosity mapping approach**
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- LXVI. **B cells produce less IL-10, IL-6 and TNF- α in myasthenia gravis**
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- LXVII. **NGLY1 mutation causes neuromotor impairment, intellectual disability, and neuropathy**
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- LXVIII. **A Drosophila Genetic Resource of Mutants to Study Mechanisms Underlying Human Genetic Diseases**
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PARMAN F. Y., TÜZÜN E., ÖZDAĞ ACARLI A. N., YILMAZ V., Project Supported by Higher Education Institutions, Kronik İnflamatuar Demiyelinizan Polinöropati Hastalarında B hücrelerine Spesifik Gen Ekspresyonlarının Belirlenmesi, 2019 - 2021

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Metrics

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Citation (WoS): 5812
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Non Academic Experience

Paris-Sud Tıp Fakültesi , Kremlin-Bicêtre Hastanesi, Louis-Ranvier Nörobiyoloji Laboratuvarı