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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, Familyal 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

- 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.
Clinical and experimental immunology, vol.215, no.1, pp.65-78, 2024 (SCI-Expanded)
- II. **Genetic landscape of congenital insensitivity to pain and hereditary sensory and autonomic**

neuropathies.

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III. Eplontersen for Hereditary Transthyretin Amyloidosis With Polyneuropathy

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JAMA-JOURNAL OF THE AMERICAN MEDICAL ASSOCIATION, 2023 (SCI-Expanded)

IV. Phenotypic features of RETREG1-related hereditary sensory autonomic neuropathy

Çakar A., Bagirova G., Durmuş H., Uyguner O., Parman Y.

Journal of the Peripheral Nervous System, vol.28, no.3, pp.351-358, 2023 (SCI-Expanded)

V. Thymoma patients with or without myasthenia gravis have increased Th17 cells, IL-17 production and ICOS expression

Cebi M., Çakar A., Erdogan E., Durmus-Tekce H., Yegen G., Ozkan B., Parman Y., Saruhan-Direskeneli G.

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VI. Characteristics of Patients with Hereditary Transthyretin Amyloidosis-Polyneuropathy (ATTRv-PN) in NEURO-TTRtransform, an Open-label Phase 3 Study of Eplontersen

Coelho T., Cruz M. W., Chao C., Parman Y., Wixner J., Weiler M., Barroso F. A., Dasgupta N. R., Jung S. W., Schneider E., et al.

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VII. 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)

VIII. 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.

Amyloid, vol.30, no.3, pp.313-320, 2023 (SCI-Expanded)

IX. Clinical and genetic profile of patients enrolled in the Transthyretin Amyloidosis Outcomes Survey (THAOS): 14-year update

Dispenzieri A., Coelho T., Conceição I., Waddington-Cruz M., Wixner J., Kristen A. V., Rapezzi C., Planté-Bordeneuve V., Gonzalez-Moreno J., Maurer M. S., et al.

Orphanet Journal of Rare Diseases, vol.17, no.1, 2022 (SCI-Expanded)

X. Disease activity in chronic inflammatory demyelinating polyneuropathy: A comparative study of clinical and skin biopsy markers

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XI. Clinical and genetic characteristics of Emery-Dreifuss muscular dystrophy patients from Turkey: 30 years longitudinal follow-up study.

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XII. 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., Samancı B., BATTALOĞLU E., Basak A. N., Bilgic B., Hanagasi H., Durmus H., et al.

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XIII. NOVEL VARIANTS BROADEN THE MUTATIONAL SPECTRUM OF HEREDITARY SENSORY AND AUTONOMIC NEUROPATHY DISORDERS

Lischka A., Eggermann K., Çakar A., Record C., Elbracht M., Hornemann T., Senderek J., Parman Y., Auer-Grumbach

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 JOURNAL OF THE PERIPHERAL NERVOUS SYSTEM, vol.27, 2022 (SCI-Expanded)
- XIV. **CHARACTERISTICS OF PATIENTS WITH HEREDITARY TRANSTHYRETIN AMYLOIDOSIS-POLYNEUROPATHY (ATTRV PN) IN NEURO-TTRANSFORM, A PHASE 3 STUDY OF EPLONTERSEN**
 Cruz M. W., Barroso F., Berk J., Chao C., Dasgupta N., Dyck P. J. B., Gertz M., Obici L., Parman Y., Weiler M., et al.
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- XV. **AN EXPLORATORY STUDY OF COGNITIVE INVOLVEMENT IN HEREDITARY ATTRV**
 Durmus H., Cakar A., Demirci H., Parman Y.
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- XVI. **Genetic pain loss disorders**
 Lischka A., Lassuthova P., cakar A., Record C. J., Van Lent J., Baets J., Dohrn M. F., Senderek J., Lampert A., Bennett D. L., et al.
 NATURE REVIEWS DISEASE PRIMERS, vol.8, no.1, 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, vol.145, pp.619-626, 2022 (SCI-Expanded)
- XVIII. **Genetics of Pain: Novel variants identified by the European Network on Inherited Sensory Neuropathies and Insensitivity to Pain (ENISNIP)**
 Lischka A., Eggermann K., Cakar A., Bocek R., Bartesaghi L., Elbracht M., Hornemann T., Senderek J., Auer-Grumbach M., Parman Y., et al.
 EUROPEAN JOURNAL OF HUMAN GENETICS, vol.30, no.SUPPL 1, pp.128, 2022 (SCI-Expanded)
- XIX. **Screening of SORD mutations in a CMT cohort expands the clinical spectrum of SORD-related neuropathy**
 Armiola-Ricaurte C., de Vriendt E., Candayan A., Asenov O., Parman Y., Chamova T., Tournev I., BATTALOĞLU E., Jordanova A.
 EUROPEAN JOURNAL OF HUMAN GENETICS, vol.30, no.SUPPL 1, pp.309-310, 2022 (SCI-Expanded)
- XX. **Bi-allelic variants in neuronal cell adhesion molecule cause a neurodevelopmental disorder characterized by developmental delay, hypotonia, neuropathy/spasticity**
 Kurolap A., Kreuder F., Gonzaga-Jauregui C., Duvdevani M. P., Harel T., Tammer L., Xin B., Bakhtiari S., Rice J., van Eyk C. L., et al.
 AMERICAN JOURNAL OF HUMAN GENETICS, vol.109, no.3, pp.518-532, 2022 (SCI-Expanded)
- XXI. **Lumbar Spinal Stenosis: A Rare Presentation of Hereditary Transthyretin Amyloidosis**
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- XXII. **Clinical and Genetic Survey for Charcot-Marie-Tooth Neuropathy Based on the Findings in Turkey, a Country with a High Rate of Consanguineous Marriages**
 Candayan A., Parman Y., BATTALOĞLU E.
 BALKAN MEDICAL JOURNAL, vol.39, no.1, pp.3-11, 2022 (SCI-Expanded)
- XXIII. **An Exploratory Study of Cognitive Involvement in Hereditary Transthyretin Amyloidosis**
 Durmus H., Cakar A., Demirci H., ALAYLİOĞLU M., GEZEN AK D., DURSUN E., Gulsen Parman Y.
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- XXIV. **Genetic Survey of Autosomal Recessive Peripheral Neuropathy Cases Unravels High Genetic Heterogeneity in a Turkish Cohort**
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- XXV. **Cerebellar ataxia, neuropathy and vestibular areflexia syndrome (Canvas) is an important cause of late-onset ataxia**
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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. **Screening of SORD mutations in a CMT cohort expands the clinical spectrum of SORD-related neuropathy**
Armirola-Ricaurte C., De Vriendt E., Candayan A., Asenov O., Parman Y., Chamova T., Tournev I., BATTALOĞLU E., Jordanova A.
JOURNAL OF THE PERIPHERAL NERVOUS SYSTEM, vol.26, no.3, pp.426, 2021 (SCI-Expanded)
- XXVIII. **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, vol.36, pp.1676-1688, 2021 (SCI-Expanded)
- XXIX. **Diaphragmatic dysfunction at the first visit to a chest diseases outpatient clinic in 500 patients with amyotrophic lateral sclerosis**
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MUSCLE & NERVE, vol.63, pp.683-689, 2021 (SCI-Expanded)
- XXX. **Late-onset TK2-Deficiency Patients from Turkey**
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- XXXI. **SOD1 Mutation: A Single Center Experience**
ÇAKAR A., DURMUŞ TEKÇE H., PARMAN F. Y.
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- XXXII. **Genotypic and phenotypic features of mutations in the HINT1 gene among Turkish patients with hereditary axonal neuropathy**
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- XXXIII. **Dysregulation of myelin synthesis and actomyosin function underlies aberrant myelin in CMT4B1 neuropathy**
Guerrero-Valero M., Grandi F., Cipriani S., Alberizzi V., Di Guardo R., Chicanne G., Sawade L., Bianchi F., Del Carro U., Curtis I. D., et al.
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- XXXIV. **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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- XXXV. **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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- XXXVI. **Revisiting the complex architecture of ALS in Turkey: Expanding genotypes, shared phenotypes, molecular networks, and a public variant database**
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- XXXVII. **Clinical and genetic aspects of hereditary spastic paraparesis in patients from Turkey**
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- XXXVIII. **A Val30Met sporadic familial amyloid polyneuropathy case with atypical presentation: upper limb onset of symptoms**
Sahin E., Cakar A., Durmus-Tekce H., Parman Y.

- ACTA NEUROLOGICA BELGICA, vol.119, no.4, pp.627-628, 2019 (SCI-Expanded)
- XXXIX. **A novel homozygous FBXO38 variant causes an early-onset distal hereditary motor neuronopathy type IID.**
Akçimen F., Vural A., Durmus H., Cakar A., Houlden H., Parman Y. G., Basak A. N.
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- XL. **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.
NEUROGENETICS, 2019 (SCI-Expanded)
- XLI. **Mutation spectrum of 260 dystrophinopathy patients from Turkey and important highlights for genetic counseling**
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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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NEUROGENETICS, vol.20, no.3, pp.117-127, 2019 (SCI-Expanded)
- XLIII. **Assessment of patients with hereditary transthyretin amyloidosis - understanding the impact of management and disease progression**
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- XLIV. **A multicenter retrospective study of charcot-marie-tooth disease type 4B (CMT4B) associated with mutations in myotubularin-related proteins (MTMRs)**
Pareyson D., Stojkovic T., Reilly M. M., Leonard-Louis S., Laura M., Blake J., Parman Y., BATTALOĞLU E., Tazir M., Bellatache M., et al.
ANNALS OF NEUROLOGY, vol.86, no.1, pp.55-67, 2019 (SCI-Expanded)
- XLV. **Familial Amyloid Polyneuropathy**
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- XLVI. **Patisiran, an RNAi Therapeutic, for Hereditary Transthyretin Amyloidosis**
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- XLVII. **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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- XLVIII. **Myophosphorylase (PYGM) mutations determined by next generation sequencing in a cohort from Turkey with McArdle disease**
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- XLIX. **MCM3AP in recessive Charcot-Marie-Tooth neuropathy and mild intellectual disability**
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- L. **Elevated IL-4 and IFN- γ Levels in Muscle Tissue of Patients with Dermatomyositis**
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- LI. **Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis**
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- LII. **Neuromuscular endplate pathology in recessive desminopathies: Lessons from man and mice**
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- LIII. **Genotypic and phenotypic presentation of transthyretin-related familial amyloid polyneuropathy (TTR-FAP) in Turkey.**
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- LIV. **Volumetric differences suggest involvement of cerebellum and brainstem in chronic migraine**
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- LV. **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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- LVI. **Optimizing the management of transthyretin familial amyloid polyneuropathy in Europe: early diagnosis and effective care.**
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- LVII. **Vocal Cord Paralysis and Hypercapnic Respiratory Failure in a Patient with Familial Amyloidotic Polyneuropathy**
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- LVIII. **Exome Sequence Analysis Suggests that Genetic Burden Contributes to Phenotypic Variability and Complex Neuropathy**
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- LIX. **Transcriptional regulator PRDM12 is essential for human pain perception**
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- LX. **Differential cytokine changes in patients with myasthenia gravis with antibodies against AChR and MuSK**
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- LXI. **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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- LXII. **The distinct genetic pattern of ALS in Turkey and novel mutations**
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- LXIII. **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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- LXIV. **NGLY1 mutation causes neuromotor impairment, intellectual disability, and neuropathy**
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- LXV. **B cells produce less IL-10, IL-6 and TNF- α in myasthenia gravis**
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- LXVI. **Unraveling the genetic landscape of autosomal recessive Charcot-Marie-Tooth neuropathies using a homozygosity mapping approach**
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- LXVII. **A Drosophila Genetic Resource of Mutants to Study Mechanisms Underlying Human Genetic Diseases**
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- LXVIII. **The association of PTPN22 R620W polymorphism is stronger with late-onset AChR-myasthenia gravis in Turkey**
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- LXIX. **BLOOD-BASED DIAGNOSTIC TESTING FOR POMPE DISEASE: CONSISTENCY BETWEEN GAA ENZYME ACTIVITY IN DRIED BLOOD SPOTS AND GAA GENE SEQUENCING RESULTS**
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Non Academic Experience

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