Mario Sabatelli

List of Publications by Year in descending order

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| # | Article | IF | CITATIONS |
|----|--|-----|-----------|
| 1 | Skin biopsy and quantitative sensory assessment in an Italian cohort of ATTRv patients with polyneuropathy and asymptomatic carriers: possible evidence of early non-length dependent denervation. Neurological Sciences, 2022, 43, 1359-1364. | 1.9 | 10 |
| 2 | Identification of genetic risk loci and prioritization of genes and pathways for myasthenia gravis: a genome-wide association study. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, . | 7.1 | 36 |
| 3 | A compound score to screen patients with hereditary transthyretin amyloidosis. Journal of Neurology, 2022, , . | 3.6 | 3 |
| 4 | Realâ€life experience with inotersen in hereditary transthyretin amyloidosis with lateâ€onset phenotype: Data from an earlyâ€access program in Italy. European Journal of Neurology, 2022, 29, 2148-2155. | 3.3 | 13 |
| 5 | Characterization of the p.L145F and p.S135N Mutations in SOD1: Impact on the Metabolism of Fibroblasts Derived from Amyotrophic Lateral Sclerosis Patients. Antioxidants, 2022, 11, 815. | 5.1 | 3 |
| 6 | MOG autoimmunity mimicking CLIPPERS syndrome: Case report and literature review. Journal of Neuroimmunology, 2022, 367, 577875. | 2.3 | 3 |
| 7 | Guillain–Barré syndrome from an emergency department view: how to better predict the outcome?. Neurological Research, 2022, , 1-5. | 1.3 | Ο |
| 8 | Generation of an induced pluripotent stem cell line (UCSCi002-A) from a patient with a variant in TARDBP gene associated with familial amyotrophic lateral sclerosis and frontotemporal dementia. Stem Cell Research, 2022, 62, 102825. | 0.7 | 1 |
| 9 | Chronic inflammatory demyelinating polyradiculoneuropathy: can a diagnosis be made in patients not fulfilling electrodiagnostic criteria?. European Journal of Neurology, 2021, 28, 620-629. | 3.3 | 15 |
| 10 | Novel variants and cellular studies on patients' primary fibroblasts support a role for NEK1 missense variants in ALS pathogenesis. Human Molecular Genetics, 2021, 30, 65-71. | 2.9 | 7 |
| 11 | Patisiran in hATTR Amyloidosis: Six-Month Latency Period before Efficacy. Brain Sciences, 2021, 11, 515. | 2.3 | 8 |
| 12 | ATTRv in Lazio-Italy: A High-Prevalence Region in a Non-Endemic Country. Genes, 2021, 12, 829. | 2.4 | 9 |
| 13 | Generation of an induced pluripotent stem cell line (CSS012-A (7672)) carrying the p.G376D heterozygous mutation in the TARDBP protein. Stem Cell Research, 2021, 53, 102356. | 0.7 | 1 |
| 14 | Targeting S100A4 with niclosamide attenuates inflammatory and profibrotic pathways in models of amyotrophic lateral sclerosis. Journal of Neuroinflammation, 2021, 18, 132. | 7.2 | 11 |
| 15 | Prolonged distal motor latency of median nerve does not improve diagnostic accuracy for CIDP. Journal of Neurology, 2021, , 1. | 3.6 | 1 |
| 16 | Ocular Involvement in Hereditary Transthyretin Amyloidosis: A Case Series Describing Novel Potential Biomarkers. Genes, 2021, 12, 927. | 2.4 | 8 |
| 17 | Renal Involvement in Hereditary Transthyretin Amyloidosis: An Italian Single-Centre Experience. Brain Sciences, 2021, 11, 980. | 2.3 | 18 |
| 18 | Thr124Met myelin protein zero mutation mimicking motor neuron disease. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2021, , 1-6. | 1.7 | 0 |

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|----|--|-----|-----------|
| 19 | Generation of an induced pluripotent stem cell line (UCSCi001-A) from a patient with early-onset amyotrophic lateral sclerosis carrying a FUS variant. Stem Cell Research, 2021, 55, 102461. | 0.7 | Ο |
| 20 | A longitudinal study defined circulating microRNAs as reliable biomarkers for disease prognosis and progression in ALS human patients. Cell Death Discovery, 2021, 7, 4. | 4.7 | 36 |
| 21 | <i>SLITRK2</i> , an X-linked modifier of the age at onset in <i>C9orf72</i> frontotemporal lobar degeneration. Brain, 2021, 144, 2798-2811. | 7.6 | 7 |
| 22 | Isolated light chain deposition disease neuropathy in a patient with multiple myeloma. Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis, 2020, 27, 67-68. | 3.0 | 1 |
| 23 | Sural nerve biopsy in peripheral neuropathies: 30-year experience from a single center. Neurological Sciences, 2020, 41, 341-346. | 1.9 | 12 |
| 24 | High-Throughput Genetic Testing in ALS: The Challenging Path of Variant Classification Considering the ACMG Guidelines. Genes, 2020, 11, 1123. | 2.4 | 15 |
| 25 | Clinical features and outcomes of the flail arm and flail leg and pure lower motor neuron MND variants: a multicentre Italian study. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 1001-1003. | 1.9 | 14 |
| 26 | Personalized Prevention in Mercury-Induced Amyotrophic Lateral Sclerosis: A Case Report. Applied Sciences (Switzerland), 2020, 10, 7839. | 2.5 | 3 |
| 27 | Epigenetic profiling of Italian patients identified methylation sites associated with hereditary transthyretin amyloidosis. Clinical Epigenetics, 2020, 12, 176. | 4.1 | 13 |
| 28 | ATTRv amyloidosis Italian Registry: clinical and epidemiological data. Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis, 2020, 27, 259-265. | 3.0 | 51 |
| 29 | hATTR Pathology: Nerve Biopsy Results from Italian Referral Centers. Brain Sciences, 2020, 10, 780. | 2.3 | 24 |
| 30 | Pathological Findings in Chronic Inflammatory Demyelinating Polyradiculoneuropathy: A Single-Center Experience. Brain Sciences, 2020, 10, 383. | 2.3 | 10 |
| 31 | <p>Diagnosis and Treatment of Hereditary Transthyretin Amyloidosis (hATTR) Polyneuropathy: Current Perspectives on Improving Patient Care</p> . Therapeutics and Clinical Risk Management, 2020, Volume 16, 109-123. | 2.0 | 78 |
| 32 | ALS skin fibroblasts reveal oxidative stress and ERK1/2-mediated cytoplasmic localization of TDP-43. Cellular Signalling, 2020, 70, 109591. | 3.6 | 18 |
| 33 | Response to: SOD1 mutations in adultâ€onset distal spinal muscular atrophy. European Journal of Neurology, 2020, 27, e74. | 3.3 | 1 |
| 34 | Antibodies to neurofascin, contactin-1, and contactin-associated protein 1 in CIDP. Neurology: Neuroimmunology and NeuroInflammation, 2020, 7, . | 6.0 | 118 |
| 35 | Relevance of diagnostic investigations in chronic inflammatory demyelinating poliradiculoneuropathy: Data from the Italian CIDP database. Journal of the Peripheral Nervous System, 2020, 25, 152-161. | 3.1 | 15 |
| 36 | SOD1 p.D12Y variant is associated with amyotrophic lateral sclerosis/distal myopathy spectrum. European Journal of Neurology, 2020, 27, 1304-1309. | 3.3 | 4 |

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|----|--|-----|-----------|
| 37 | Gastrointestinal Manifestations in Hereditary Transthyretin Amyloidosis: a Single-Centre Experience. Journal of Gastrointestinal and Liver Diseases, 2020, 29, 339-343. | 0.9 | 10 |
| 38 | Small Fibre Involvement in Multifocal Motor Neuropathy Explored with Sudoscan: A Single-Centre Experience. Diagnostics, 2020, 10, 755. | 2.6 | 2 |
| 39 | The S100A4 Transcriptional Inhibitor Niclosamide Reduces Pro-Inflammatory and Migratory Phenotypes of Microglia: Implications for Amyotrophic Lateral Sclerosis. Cells, 2019, 8, 1261. | 4.1 | 24 |
| 40 | Histamine beyond its effects on allergy: Potential therapeutic benefits for the treatment of Amyotrophic Lateral Sclerosis (ALS). , 2019, 202, 120-131. | | 19 |
| 41 | Comparative Analysis of C9orf72 and Sporadic Disease in a Large Multicenter ALS Population: The Effect of Male Sex on Survival of C9orf72 Positive Patients. Frontiers in Neuroscience, 2019, 13, 485. | 2.8 | 35 |
| 42 | Proteostasis and ALS: protocol for a phase II, randomised, double-blind, placebo-controlled, multicentre clinical trial for colchicine in ALS (Co-ALS). BMJ Open, 2019, 9, e028486. | 1.9 | 44 |
| 43 | A multicenter retrospective study of charcotâ€marieâ€tooth disease type 4B (CMT4B) associated with mutations in myotubularinâ€related proteins (MTMRs). Annals of Neurology, 2019, 86, 55-67. | 5.3 | 35 |
| 44 | Coexistence of variants in TBK1 and in other ALS-related genes elucidates an oligogenic model of pathogenesis in sporadic ALS. Neurobiology of Aging, 2019, 84, 239.e9-239.e14. | 3.1 | 21 |
| 45 | Relations between C9orf72 expansion size in blood, age at onset, age at collection and transmission across generations in patients and presymptomatic carriers. Neurobiology of Aging, 2019, 74, 234.e1-234.e8. | 3.1 | 38 |
| 46 | ATXN1 intermediate-length polyglutamine expansions are associated with amyotrophic lateral sclerosis. Neurobiology of Aging, 2018, 64, 157.e1-157.e5. | 3.1 | 34 |
| 47 | Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6. | 8.1 | 517 |
| 48 | Sudoscan in the evaluation and follow-up of patients and carriers with TTR mutations: experience from an Italian Centre. Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis, 2018, 25, 242-246. | 3.0 | 28 |
| 49 | PERIPHERAL NERVOUS SYSTEM INVOLVEMENT IN LYMPHOPROLIFERATIVE DISORDERS. Mediterranean Journal of Hematology and Infectious Diseases, 2018, 10, e2018057. | 1.3 | 1 |
| 50 | Generation and characterization of a human iPSC line from an ALS patient carrying the Q66K-MATR3 mutation. Stem Cell Research, 2018, 33, 146-150. | 0.7 | 3 |
| 51 | Elevated Levels of Selenium Species in Cerebrospinal Fluid of Amyotrophic Lateral Sclerosis Patients with Disease-Associated Gene Mutations. Neurodegenerative Diseases, 2017, 17, 171-180. | 1.4 | 46 |
| 52 | Non-coding variants contribute to the clinical heterogeneity of TTR amyloidosis. European Journal of Human Genetics, 2017, 25, 1055-1060. | 2.8 | 23 |
| 53 | Comorbidity of dementia with amyotrophic lateral sclerosis (ALS): insights from a large multicenter Italian cohort. Journal of Neurology, 2017, 264, 2224-2231. | 3.6 | 19 |
| 54 | Potential therapeutic targets for ALS: MIR206, MIR208b and MIR499 are modulated during disease progression in the skeletal muscle of patients. Scientific Reports, 2017, 7, 9538. | 3.3 | 48 |

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|----|--|-----|-----------|
| 55 | Factors predicting survival in ALS: a multicenter Italian study. Journal of Neurology, 2017, 264, 54-63. | 3.6 | 96 |
| 56 | Matrin 3 variants are frequent in Italian ALS patients. Neurobiology of Aging, 2017, 49, 218.e1-218.e7. | 3.1 | 35 |
| 57 | Recurrent miller fisher: a new case report and a literature review. Clinica Terapeutica, 2017, 168, e208-e213. | 0.3 | 5 |
| 58 | Nerve ultrasound in CMT2E/CMT1F due to NEFL mutation: Confirmation of an axonal pathology. Clinical Neurophysiology, 2016, 127, 2990-2991. | 1.5 | 6 |
| 59 | TBK1 is associated with ALS and ALS-FTD in Sardinian patients. Neurobiology of Aging, 2016, 43, 180.e1-180.e5. | 3.1 | 40 |
| 60 | Monitoring effectiveness and safety of Tafamidis in transthyretin amyloidosis in Italy: a longitudinal multicenter study in a non-endemic area. Journal of Neurology, 2016, 263, 916-924. | 3.6 | 76 |
| 61 | Start codon mutation of GYG1 causing late-onset polyglucosan body myopathy with nemaline rods. Journal of Neurology, 2016, 263, 2133-2135. | 3.6 | 17 |
| 62 | New ALSâ€Related Genes Expand the <i>Spectrum Paradigm</i> of Amyotrophic Lateral Sclerosis. Brain Pathology, 2016, 26, 266-275. | 4.1 | 26 |
| 63 | ATNX2 is not a regulatory gene in Italian amyotrophic lateral sclerosis patients with C9ORF72 GGGGCC expansion. Neurobiology of Aging, 2016, 39, 218.e5-218.e8. | 3.1 | 6 |
| 64 | Charcot-Marie-Tooth type 2 and distal hereditary motor neuropathy: Clinical, neurophysiological and genetic findings from a single-centre experience. Clinical Neurology and Neurosurgery, 2016, 144, 67-71. | 1.4 | 18 |
| 65 | Nerve ultrasound in patients with CMT1C: Description of 3 cases. Muscle and Nerve, 2015, 51, 781-782. | 2.2 | 7 |
| 66 | Flow Cytofluorimetric Analysis of Anti-LRP4 (LDL Receptor-Related Protein 4) Autoantibodies in Italian Patients with Myasthenia Gravis. PLoS ONE, 2015, 10, e0135378. | 2.5 | 30 |
| 67 | Neuromyelitis optica spectrum disorder as a paraneoplastic manifestation of lung adenocarcinoma expressing aquaporin-4. Multiple Sclerosis Journal, 2015, 21, 791-794. | 3.0 | 28 |
| 68 | Admission neurophysiological abnormalities in Guillain–Barré syndrome: A single-center experience. Clinical Neurology and Neurosurgery, 2015, 135, 6-10. | 1.4 | 15 |
| 69 | â€~Behr syndrome' with OPA1 compound heterozygote mutations. Brain, 2015, 138, e321-e321. | 7.6 | 50 |
| 70 | Letter: faecal microbiota transplantation in combination with fidaxomicin to treat severe complicated recurrent <i>Clostridium difficile</i> infection. Alimentary Pharmacology and Therapeutics, 2015, 42, 1030-1030. | 3.7 | 11 |
| 71 | Nerve ultrasound findings in neuropathy associated with antiâ€myelinâ€associated glycoprotein antibodies. European Journal of Neurology, 2015, 22, 193-202. | 3.3 | 34 |
| 72 | Skin Changes in POEMS Syndrome. European Neurology, 2015, 73, 112-112. | 1.4 | 0 |

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|----|--|------|-----------|
| 73 | Primary fibroblasts cultures reveal TDP-43 abnormalities in amyotrophic lateral sclerosis patients with and without SOD1 mutations. Neurobiology of Aging, 2015, 36, 2005.e5-2005.e13. | 3.1 | 42 |
| 74 | Distinct lymphocytes subsets in IgM-related neuropathy: clinical-immunological correlations. Neurological Sciences, 2015, 36, 303-308. | 1.9 | 2 |
| 75 | Frequency and time to relapse after discontinuing 6-month therapy with IVIg or pulsed methylprednisolone in CIDP. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 729-734. | 1.9 | 70 |
| 76 | HFE p.H63D polymorphism does not influence ALS phenotype and survival. Neurobiology of Aging, 2015, 36, 2906.e7-2906.e11. | 3.1 | 8 |
| 77 | A Genome-Wide Association Study of Myasthenia Gravis. JAMA Neurology, 2015, 72, 396. | 9.0 | 139 |
| 78 | CHCH10 mutations in an Italian cohort of familial and sporadic amyotrophic lateral sclerosis patients. Neurobiology of Aging, 2015, 36, 1767.e3-1767.e6. | 3.1 | 44 |
| 79 | 'White Nails'. European Neurology, 2015, 73, 89-89. | 1.4 | 2 |
| 80 | <i>ATXN2</i> polyQ intermediate repeats are a modifier of ALS survival. Neurology, 2015, 84, 251-258. | 1.1 | 52 |
| 81 | Primary multifocal lymphoma of peripheral nervous system: Case report and review of the literature. Muscle and Nerve, 2014, 50, 1016-1022. | 2.2 | 30 |
| 82 | Ultrasound evaluation in transthyretinâ€related amyloid neuropathy. Muscle and Nerve, 2014, 50, 372-376. | 2.2 | 32 |
| 83 | Clinical, neurophysiological and pathological findings of HNPP patients with 17p12 deletion: A single-centre experience. Journal of the Neurological Sciences, 2014, 341, 46-50. | 0.6 | 32 |
| 84 | Mutations in the Matrin 3 gene cause familial amyotrophic lateral sclerosis. Nature Neuroscience, 2014, 17, 664-666. | 14.8 | 398 |
| 85 | Genetic counselling in ALS: facts, uncertainties and clinical suggestions. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 478-485. | 1.9 | 99 |
| 86 | Heterogeneity of root and nerve ultrasound pattern in CIDP patients. Clinical Neurophysiology, 2014, 125, 160-165. | 1.5 | 142 |
| 87 | Mutations in the CHCHD10 gene are a common cause of familial amyotrophic lateral sclerosis. Brain, 2014, 137, e311-e311. | 7.6 | 112 |
| 88 | An ALS-associated mutation in the FUS 3′-UTR disrupts a microRNA–FUS regulatory circuitry. Nature Communications, 2014, 5, 4335. | 12.8 | 102 |
| 89 | Tuberculous nephritis accompanying neuromyelitis optica: causal or coincidental association?. Journal of Neurology, 2014, 261, 1028-1030. | 3.6 | 4 |
| 90 | Clinical, electrophysiological and pathological findings in a patient with Charcot–Marie–Tooth disease 4D caused by the NDRG1 Lom mutation. Journal of the Neurological Sciences, 2014, 345, 271-273. | 0.6 | 6 |

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|-----|--|-----|-----------|
| 91 | TTR-related amyloid neuropathy: clinical, electrophysiological and pathological findings in 15 unrelated patients. Neurological Sciences, 2013, 34, 1057-1063. | 1.9 | 43 |
| 92 | Clinical–neurophysiological correlations in a series of patients with IgM-related neuropathy. Clinical Neurophysiology, 2013, 124, 1899-1903. | 1.5 | 17 |
| 93 | Clinical and genetic heterogeneity ofÂamyotrophic lateral sclerosis. Clinical Genetics, 2013, 83, 408-416. | 2.0 | 92 |
| 94 | Influence of comorbidities on the phenotype of patients affected by Charcot–Marie–Tooth neuropathy type 1A. Neuromuscular Disorders, 2013, 23, 902-906. | 0.6 | 15 |
| 95 | Mutations in the $3\hat{a}\in^2$ untranslated region of FUS causing FUS overexpression are associated with amyotrophic lateral sclerosis. Human Molecular Genetics, 2013, 22, 4748-4755. | 2.9 | 94 |
| 96 | Inherited neuropathies and deafness caused by a PMP22 point mutation: a case report and a review of the literature. Neurological Sciences, 2013, 34, 1705-1707. | 1.9 | 6 |
| 97 | A novel compound heterozygous <i>ALS2</i> mutation in two Italian siblings with juvenile amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 470-472. | 1.7 | 12 |
| 98 | Frontotemporal dementia, Parkinsonism and lower motor neuron involvement in a patient with C9ORF72 expansion. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 66-69. | 1.7 | 13 |
| 99 | A novel homozygous mutation in the <i><scp>MTMR2</scp></i> gene in two siblings with â€ ⁻ hypermyelinating neuropathy'. Journal of the Peripheral Nervous System, 2013, 18, 192-194. | 3.1 | 16 |
| 100 | MRI Neurography Findings in Patients with Idiopathic Brachial Plexopathy: Correlations with Clinical-neurophysiological Data in Eight Consecutive Cases. Internal Medicine, 2013, 52, 2031-2039. | 0.7 | 7 |
| 101 | Mitochondrial Network Genes in the Skeletal Muscle of Amyotrophic Lateral Sclerosis Patients. PLoS ONE, 2013, 8, e57739. | 2.5 | 42 |
| 102 | Restless Leg Syndrome in Different Types of Demyelinating Neuropathies: A Single-Center Pilot Study. Journal of Clinical Sleep Medicine, 2013, 09, 945-949. | 2.6 | 12 |
| 103 | Founder effect hypothesis of D11Y SOD1 mutation in Italian amyotrophic lateral sclerosis patients. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2012, 13, 241-242. | 2.1 | 4 |
| 104 | Classification of familial amyotrophic lateral sclerosis by family history: effects on frequency of genes mutation. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, 1201-1203. | 1.9 | 22 |
| 105 | AL amyloid neuropathy mimicking a chronic inflammatory demyelinating polyneuropathy. Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis, 2012, 19, 53-55. | 3.0 | 20 |
| 106 | Teaching Neurolmages: Peroneal intraneural ganglion cyst: A rare cause of drop foot in a child. Neurology, 2012, 78, e46-e47. | 1.1 | 13 |
| 107 | Usefulness of F-18 FDG PET/CT in the Follow-up of POEMS Syndrome After Autologous Peripheral Blood Stem Cell Transplantation. Clinical Nuclear Medicine, 2012, 37, 181-183. | 1.3 | 14 |
| 108 | Nutritional and metabolic support in patients with amyotrophic lateral sclerosis. Nutrition, 2012, 28, 959-966. | 2.4 | 48 |

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|-----|---|------|-----------|
| 109 | Sural nerve pathology in ALS patients: a single-centre experience. Neurological Sciences, 2012, 33, 1095-1099. | 1.9 | 17 |
| 110 | Replication of association of CHRNA4 rare variants with sporadic amyotrophic lateral sclerosis: The Italian multicentre study. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2012, 13, 580-584. | 2.1 | 7 |
| 111 | C9ORF72 hexanucleotide repeat expansions in the Italian sporadic ALS population. Neurobiology of Aging, 2012, 33, 1848.e15-1848.e20. | 3.1 | 76 |
| 112 | P525L FUS mutation is consistently associated with a severe form of juvenile Amyotrophic Lateral Sclerosis. Neuromuscular Disorders, 2012, 22, 73-75. | 0.6 | 124 |
| 113 | Clinical characteristics of patients with familial amyotrophic lateral sclerosis carrying the pathogenic GGGGCC hexanucleotide repeat expansion of C9ORF72. Brain, 2012, 135, 784-793. | 7.6 | 182 |
| 114 | Peripheral neuropathy and 46XY gonadal dysgenesis: Confirmation of a heterogeneous entity. Clinical Neurology and Neurosurgery, 2012, 114, 748-750. | 1.4 | 2 |
| 115 | Cranial botulism. Neuromuscular Disorders, 2012, 22, 995-996. | 0.6 | 2 |
| 116 | Clinical and pathological heterogeneity in a series of 31 patients with IgM-related neuropathy. Journal of the Neurological Sciences, 2012, 319, 75-80. | 0.6 | 18 |
| 117 | Contribution of major amyotrophic lateral sclerosis genes to the etiology of sporadic disease. Neurology, 2012, 79, 66-72. | 1.1 | 99 |
| 118 | Efficacy of lenalidomide plus dexamethasone for POEMS syndrome relapsed after autologous peripheral stem ell transplantation. American Journal of Hematology, 2012, 87, 641-642. | 4.1 | 24 |
| 119 | Frequency of the C9orf72 hexanucleotide repeat expansion in patients with amyotrophic lateral sclerosis and frontotemporal dementia: a cross-sectional study. Lancet Neurology, The, 2012, 11, 323-330. | 10.2 | 1,039 |
| 120 | Intravenous immunoglobulin versus intravenous methylprednisolone for chronic inflammatory demyelinating polyradiculoneuropathy: a randomised controlled trial. Lancet Neurology, The, 2012, 11, 493-502. | 10.2 | 185 |
| 121 | A novel L67P SOD1 mutation in an Italian ALS patient. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2011, 12, 150-152. | 2.1 | 11 |
| 122 | Rituximab in patients with chronic inflammatory demyelinating polyradiculoneuropathy: a report of 13 cases and review of the literature. Journal of Neurology, Neurosurgery and Psychiatry, 2011, 82, 306-308. | 1.9 | 106 |
| 123 | Clinical, electrophysiological and pathological findings of a patient with CMT2 due to the p.Ala738Val mitofusin 2 mutation. Journal of the Neurological Sciences, 2011, 307, 168-170. | 0.6 | 8 |
| 124 | D11Y SOD1 mutation and benign ALS: A consistent genotype-phenotype correlation. Journal of the Neurological Sciences, 2011, 309, 31-33. | 0.6 | 12 |
| 125 | FUS mutations in sporadic amyotrophic lateral sclerosis. Neurobiology of Aging, 2011, 32, 550.e1-550.e4. | 3.1 | 79 |
| 126 | SOD1 G93D sporadic amyotrophic lateral sclerosis (SALS) patient with rapid progression and concomitant novel ANG variant. Neurobiology of Aging, 2011, 32, 1924.e15-1924.e18. | 3.1 | 32 |

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|-----|--|-----|-----------|
| 127 | Exome Sequencing Reveals VCP Mutations as a Cause of Familial ALS. Neuron, 2011, 69, 397. | 8.1 | 7 |
| 128 | A Hexanucleotide Repeat Expansion in C9ORF72 Is the Cause of Chromosome 9p21-Linked ALS-FTD. Neuron, 2011, 72, 257-268. | 8.1 | 3,833 |
| 129 | Abnormal vascular smooth muscle cell proliferation in sural nerve biopsy from a patient with sensorimotor axonal neuropathy. Neuropathology, 2011, 31, 197-198. | 1.2 | 1 |
| 130 | Immunosuppressive treatment in refractory chronic inflammatory demyelinating polyradiculoneuropathy. A nationwide retrospective analysis. European Journal of Neurology, 2011, 18, 1417-1421. | 3.3 | 71 |
| 131 | Mutant human β4 subunit identified in amyotrophic lateral sclerosis patients impairs nicotinic receptor function. Pflugers Archiv European Journal of Physiology, 2011, 461, 225-233. | 2.8 | 8 |
| 132 | Progressive ascending myelopathy: atypical forms of multiple sclerosis or what else?. Journal of Neurology, 2011, 258, 1965-1970. | 3.6 | 3 |
| 133 | Botulinum toxin A versus B in sialorrhea: A prospective, randomized, double-blind, crossover pilot study in patients with amyotrophic lateral sclerosis or Parkinson's disease. Movement Disorders, 2011, 26, 313-319. | 3.9 | 111 |
| 134 | Repeated courses of granulocyte colonyâ€stimulating factor in amyotrophic lateral sclerosis: Clinical and biological results from a prospective multicenter study. Muscle and Nerve, 2011, 43, 189-195. | 2.2 | 64 |
| 135 | A novel <i>GJB1</i> mutation in an Italian patient with Charcot–Marie–Tooth disease and pyramidal signs. Muscle and Nerve, 2011, 44, 613-615. | 2.2 | 2 |
| 136 | Uncovering amyotrophic lateral sclerosis phenotypes: Clinical features and long-term follow-up of upper motor neuron-dominant ALS. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2011, 12, 278-282. | 2.1 | 32 |
| 137 | Teaching Neuro <i>Images</i> : Cochleitis. Neurology, 2011, 77, e109. | 1.1 | 1 |
| 138 | Neuropathy with Predominant Small Fiber Involvement Associated with Abnormal Anti-MAG Titer. Internal Medicine, 2010, 49, 2627-2629. | 0.7 | 10 |
| 139 | TEACHING NEURO <i>IMAGES</i> : THE FULL-BLOWN NEUROIMAGING OF WERNICKE ENCEPHALOPATHY. Neurology, 2010, 74, 527-528. | 1.1 | 3 |
| 140 | Demyelinating encephalomyeloradiculitis with Balò-like lesions. Journal of Neurology, 2010, 257, 1566-1567. | 3.6 | 2 |
| 141 | Epstein-Barr virus antibodies in serum and cerebrospinal fluid from Multiple sclerosis, Chronic Inflammatory Demyelinating Polyradiculoneuropathy and Amyotrophic Lateral Sclerosis. Journal of Neuroimmunology, 2010, 225, 149-152. | 2.3 | 33 |
| 142 | Long-term motor cortex stimulation for amyotrophic lateral sclerosis. Brain Stimulation, 2010, 3, 22-27. | 1.6 | 20 |
| 143 | <i>SEIPIN</i> S90L Mutation in an Italian family with CMT2/dHMN and pyramidal signs. Muscle and Nerve, 2010, 42, 448-451. | 2.2 | 16 |
| 144 | A nationwide retrospective analysis on the effect of immune therapies in patients with chronic inflammatory demyelinating polyradiculoneuropathy. European Journal of Neurology, 2010, 17, 289-294. | 3.3 | 115 |

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|-----|--|-----|-----------|
| 145 | Posterior ischaemic myelopathy associated with cocaine abuse. Internal Medicine Journal, 2010, 40, 732-733. | 0.8 | 4 |
| 146 | Lithium carbonate in amyotrophic lateral sclerosis. Neurology, 2010, 75, 619-625. | 1.1 | 90 |
| 147 | Gadolinium enhancement of the lumbar leptomeninges and roots in a case of ALS. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2010, 11, 412-413. | 2.1 | 8 |
| 148 | Triple A syndrome: A novel compound heterozygous mutation in the AAAS gene in an Italian patient without adrenal insufficiency. Journal of the Neurological Sciences, 2010, 290, 150-152. | 0.6 | 15 |
| 149 | Light chain deposition in peripheral nerve as a cause of mononeuritis multiplex in Waldenström's macroglobulinaemia. Journal of the Neurological Sciences, 2010, 291, 89-91. | 0.6 | 23 |
| 150 | A novel HSPB1 mutation in an Italian patient with CMT2/dHMN phenotype. Journal of the Neurological Sciences, 2010, 298, 114-117. | 0.6 | 42 |
| 151 | A case of CMT 1B due to Val 102/fs null mutation of the MPZ gene presenting as hyperCKemia. Clinical Neurology and Neurosurgery, 2010, 112, 794-797. | 1.4 | 13 |
| 152 | Exome Sequencing Reveals VCP Mutations as a Cause of Familial ALS. Neuron, 2010, 68, 857-864. | 8.1 | 1,100 |
| 153 | An Italian case of hereditary myopathy with early respiratory failure (HMERF) not associated with the titin kinase domain R279W mutation. Neuromuscular Disorders, 2010, 20, 730-734. | 0.6 | 15 |
| 154 | Consistent bone marrow-derived cell mobilization following repeated short courses of granulocyte–colony-stimulating factor in patients with amyotrophic lateral sclerosis: results from a multicenter prospective trial. Cytotherapy, 2010, 12, 50-59. | 0.7 | 36 |
| 155 | Teaching Neuro <i>Image</i> : MRI of diabetic lumbar plexopathy treated with local steroid injection. Neurology, 2009, 72, e32-3. | 1.1 | 3 |
| 156 | A two-stage genome-wide association study of sporadic amyotrophic lateral sclerosis. Human Molecular Genetics, 2009, 18, 1524-1532. | 2.9 | 106 |
| 157 | Teaching Neuro <i>Images</i> : The full-blown neuroimaging of Wernicke encephalopathy. Neurology, 2009, 72, e115. | 1.1 | 8 |
| 158 | NATURAL HISTORY OF YOUNG-ADULT AMYOTROPHIC LATERAL SCLEROSIS. Neurology, 2009, 73, 648-650. | 1.1 | 0 |
| 159 | NEUROLOGIC IMPROVEMENT AFTER PERIPHERAL BLOOD STEM CELL TRANSPLANTATION IN POEMS. Neurology, 2009, 73, 1165-1166. | 1.1 | 4 |
| 160 | Rare missense variants of neuronal nicotinic acetylcholine receptor altering receptor function are associated with sporadic amyotrophic lateral sclerosis. Human Molecular Genetics, 2009, 18, 3997-4006. | 2.9 | 42 |
| 161 | Ultrasound visualization of nerve morphological alteration at the site of conduction block. Muscle and Nerve, 2009, 40, 1068-1070. | 2.2 | 54 |
| 162 | Heterozygous SOD1 D90A mutation presenting as slowly progressive predominant upper motor neuron amyotrophic lateral sclerosis. Neurological Sciences, 2009, 30, 517-520. | 1.9 | 21 |

| # | Article | IF | CITATIONS |
|-----|--|-----|-----------|
| 163 | pSTAT1, pSTAT3, and Tâ€bet as markers of disease activity in chronic inflammatory demyelinating polyradiculoneuropathy. Journal of the Peripheral Nervous System, 2009, 14, 107-117. | 3.1 | 31 |
| 164 | Two Italian kindreds with familial amyotrophic lateral sclerosis due to FUS mutation. Neurobiology of Aging, 2009, 30, 1272-1275. | 3.1 | 128 |
| 165 | Motor cortex stimulation for ALS: A double blind placebo-controlled study. Neuroscience Letters, 2009, 464, 18-21. | 2.1 | 33 |
| 166 | Progressive multifocal leukoencephalopathy in a patient with Franklin disease and hypogammaglobulinemia. Journal of the Neurological Sciences, 2009, 284, 203-204. | 0.6 | 9 |
| 167 | SOD1 G93D mutation presenting as paucisymptomatic amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2009, 10, 479-482. | 2.1 | 12 |
| 168 | Retinal Detachment with an Unusual Shape. Internal Medicine, 2009, 48, 1777-1778. | 0.7 | 0 |
| 169 | An Italian family with inclusionâ€body myopathy and frontotemporal dementia due to mutation in the <i>VCP</i> gene. Muscle and Nerve, 2008, 37, 111-114. | 2.2 | 44 |
| 170 | A new singleâ€nucleotide deletion of PMP22 in an HNPP family without recurrent palsies. Muscle and Nerve, 2008, 38, 1060-1064. | 2.2 | 14 |
| 171 | Early diagnosis followed by front-line autologous peripheral blood stem cell transplantation for patients affected by POEMS syndrome. Leukemia Research, 2008, 32, 1309-1312. | 0.8 | 30 |
| 172 | Natural history of young-adult amyotrophic lateral sclerosis. Neurology, 2008, 71, 876-881. | 1.1 | 81 |
| 173 | Chronic GM2 gangliosidosis type Sandhoff associated with a novel missense HEXB gene mutation causing a double pathogenic effect. Molecular Genetics and Metabolism, 2007, 91, 111-114. | 1.1 | 14 |
| 174 | Botulinum toxin B ultrasound-guided injections for sialorrhea in amyotrophic lateral sclerosis and Parkinson's disease. Parkinsonism and Related Disorders, 2007, 13, 299-303. | 2.2 | 55 |
| 175 | Repetitive transcranial magnetic stimulation for ALS. Neuroscience Letters, 2006, 408, 135-140. | 2.1 | 43 |
| 176 | Monophasic demyelinating disease of the central nervous system associated with Hepatitis A infection. Journal of Neurology, 2006, 253, 944-945. | 3.6 | 5 |
| 177 | Intravenous Immunoglobulin Treatment in Autoimmune Neurological Disorders—Effects on Quality of Life. Human Immunology, 2005, 66, 417-421. | 2.4 | 9 |
| 178 | Is carpal tunnel syndrome surgery useful in patients with diabetes or autoimmune polyneuropathies?. Journal of the Peripheral Nervous System, 2004, 9, 109-109. | 3.1 | 0 |
| 179 | Occurrence of nerve entrapment lesion in chronic inflammatory demyelinating polyneuropathy. Clinical Neurophysiology, 2004, 115, 1140-1144. | 1.5 | 15 |
| 180 | Motor cortex stimulation for amyotrophic lateral sclerosis. Time for a therapeutic trial?. Clinical Neurophysiology, 2004, 115, 1479-1485. | 1.5 | 38 |

| # | Article | IF | CITATIONS |
|-----|---|-----|-----------|
| 181 | Multinevritis of cranial nerves following inhalation of toxins. Neurological Research, 2003, 25, 208-210. | 1.3 | 0 |
| 182 | Peripheral neuropathy with hypomyelinating features in adult-onset Krabbe's disease. Neuromuscular Disorders, 2002, 12, 386-391. | 0.6 | 34 |
| 183 | Hypermyelinating neuropathy, mental retardation and epilepsy in a case of merosin deficiency. Neuromuscular Disorders, 2002, 12, 392-398. | 0.6 | 46 |
| 184 | A useful electrophysiological test for diagnosis of minimal conduction block. Clinical Neurophysiology, 2001, 112, 1041-1048. | 1.5 | 15 |
| 185 | Reply to Dr van Dijk. Clinical Neurophysiology, 2001, 112, 2164-2165. | 1.5 | Ο |
| 186 | Pure motor chronic inflammatory demyelinating polyneuropathy. Journal of Neurology, 2001, 248, 772-777. | 3.6 | 102 |
| 187 | A Useful Electrophysiologic Test For Diagnosis Of Minimal Conduction Block. Journal of the Peripheral Nervous System, 2001, 6, 54-55. | 3.1 | Ο |
| 188 | Autosornal recessive hereditary motor and sensory neuropathy with focally folded myelin sheaths. Neurology, 1996, 46, 1318-1318. | 1.1 | 121 |
| 189 | Chronic progressive steroid responsive axonal polyneuropathy: A CIDP variant or a primary axonal disorder?. , 1996, 19, 365-371. | | 37 |
| 190 | Differential electrophysiological features of neuropathies associated with 17p11.2 deletion and duplication. Muscle and Nerve, 1995, 18, 628-635. | 2.2 | 60 |
| 191 | Interferon-alpha may benefit steroid unresponsive chronic inflammatory demyelinating polyneuropathy Journal of Neurology, Neurosurgery and Psychiatry, 1995, 58, 638-639. | 1.9 | 36 |
| 192 | Autosomal recessive hypermyelinating neuropathy. Acta Neuropathologica, 1994, 87, 337-342. | 7.7 | 17 |
| 193 | Ophthalmoplegia, demyelinating neuropathy, leukoencephalopathy, myopathy, and gastrointestinal dysfunction with multiple deletions of mitochondrial DNA: A mitochondrial multisystem disorder in search of a name. Muscle and Nerve, 1994, 17, 667-674. | 2.2 | 46 |
| 194 | Hereditary motor and sensory neuropathy with calf hypertrophy is associated with 17p 11.2 duplication. Annals of Neurology, 1994, 35, 552-558. | 5.3 | 29 |
| 195 | Sensitivity and specificity of diagnostic criteria for conduction block in chronic inflammatory demyelinating polyneuropathy. Electroencephalography and Clinical Neurophysiology - Evoked Potentials, 1993, 89, 161-169. | 2.0 | 42 |
| 196 | Peripheral neuropathy with giant axons and cardiomyopathy associated with desmin type intermediate filaments in skeletal muscle. Journal of the Neurological Sciences, 1992, 109, 1-10. | 0.6 | 60 |
| 197 | Giant axonal neuropathy: report on a case with focal fiber loss. Acta Neuropathologica, 1992, 83, 543-546. | 7.7 | 20 |
| 198 | Acute axonal idiopathic polyneuropathy: A Guillain-Barré syndrome variant?. Italian Journal of Neurological Sciences, 1992, 13, 481-486. | 0.1 | 6 |

| # | Article | IF | CITATIONS |
|-----|--|-----|-----------|
| 199 | Peripheral sensory-motor polyneuropathy, pigmentary retinopathy, and fatal cardiomyopathy in long-chain 3-hydroxy-acyl-CoA dehydrogenase deficiency. European Journal of Pediatrics, 1992, 151, 121-126. | 2.7 | 57 |
| 200 | Neuromyopathy and restrictive cardiomyopathy with accumulation of intermediate filaments: a clinical, morphological and biochemical study. Acta Neuropathologica, 1991, 81, 632-640. | 7.7 | 51 |
| 201 | Genome-Wide Analyses Identify KIF5A as a Novel ALS Gene. SSRN Electronic Journal, 0, , . | 0.4 | 4 |