Mario Sabatelli

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/782631/publications.pdf

Version: 2024-02-01

201	13,103	53660	25716
papers	citations	45 h-index	108 g-index

205	205	205	12558
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A Hexanucleotide Repeat Expansion in C9ORF72 Is the Cause of Chromosome 9p21-Linked ALS-FTD. Neuron, 2011, 72, 257-268.	3.8	3,833
2	Exome Sequencing Reveals VCP Mutations as a Cause of Familial ALS. Neuron, 2010, 68, 857-864.	3.8	1,100
3	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.	4.9	1,039
4	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	3.8	517
5	Mutations in the Matrin 3 gene cause familial amyotrophic lateral sclerosis. Nature Neuroscience, 2014, 17, 664-666.	7.1	398
6	Intravenous immunoglobulin versus intravenous methylprednisolone for chronic inflammatory demyelinating polyradiculoneuropathy: a randomised controlled trial. Lancet Neurology, The, 2012, 11, 493-502.	4.9	185
7	Clinical characteristics of patients with familial amyotrophic lateral sclerosis carrying the pathogenic GGGGCC hexanucleotide repeat expansion of C9ORF72. Brain, 2012, 135, 784-793.	3.7	182
8	Heterogeneity of root and nerve ultrasound pattern in CIDP patients. Clinical Neurophysiology, 2014, 125, 160-165.	0.7	142
9	A Genome-Wide Association Study of Myasthenia Gravis. JAMA Neurology, 2015, 72, 396.	4.5	139
10	Two Italian kindreds with familial amyotrophic lateral sclerosis due to FUS mutation. Neurobiology of Aging, 2009, 30, 1272-1275.	1.5	128
11	P525L FUS mutation is consistently associated with a severe form of juvenile Amyotrophic Lateral Sclerosis. Neuromuscular Disorders, 2012, 22, 73-75.	0.3	124
12	Autosornal recessive hereditary motor and sensory neuropathy with focally folded myelin sheaths. Neurology, 1996, 46, 1318-1318.	1.5	121
13	Antibodies to neurofascin, contactin-1, and contactin-associated protein 1 in CIDP. Neurology: Neuroimmunology and NeuroInflammation, 2020, 7, .	3.1	118
14	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.	1.7	115
15	Mutations in the CHCHD10 gene are a common cause of familial amyotrophic lateral sclerosis. Brain, 2014, 137, e311-e311.	3.7	112
16	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.	2.2	111
17	A two-stage genome-wide association study of sporadic amyotrophic lateral sclerosis. Human Molecular Genetics, 2009, 18, 1524-1532.	1.4	106
18	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.	0.9	106

#	Article	IF	Citations
19	Pure motor chronic inflammatory demyelinating polyneuropathy. Journal of Neurology, 2001, 248, 772-777.	1.8	102
20	An ALS-associated mutation in the FUS 3â€²-UTR disrupts a microRNAâ€“FUS regulatory circuitry. Nature Communications, 2014, 5, 4335.	5.8	102
21	Contribution of major amyotrophic lateral sclerosis genes to the etiology of sporadic disease. Neurology, 2012, 79, 66-72.	1.5	99
22	Genetic counselling in ALS: facts, uncertainties and clinical suggestions. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 478-485.	0.9	99
23	Factors predicting survival in ALS: a multicenter Italian study. Journal of Neurology, 2017, 264, 54-63.	1.8	96
24	Mutations in the 3â€² untranslated region of FUS causing FUS overexpression are associated with amyotrophic lateral sclerosis. Human Molecular Genetics, 2013, 22, 4748-4755.	1.4	94
25	Clinical and genetic heterogeneity ofÂamyotrophic lateral sclerosis. Clinical Genetics, 2013, 83, 408-416.	1.0	92
26	Lithium carbonate in amyotrophic lateral sclerosis. Neurology, 2010, 75, 619-625.	1.5	90
27	Natural history of young-adult amyotrophic lateral sclerosis. Neurology, 2008, 71, 876-881.	1.5	81
28	FUS mutations in sporadic amyotrophic lateral sclerosis. Neurobiology of Aging, 2011, 32, 550.e1-550.e4.	1.5	79
29	Diagnosis and Treatment of Hereditary Transthyretin Amyloidosis (hATTR) Polyneuropathy: Current Perspectives on Improving Patient Care . Therapeutics and Clinical Risk Management, 2020, Volume 16, 109-123.	0.9	78
30	C9ORF72 hexanucleotide repeat expansions in the Italian sporadic ALS population. Neurobiology of Aging, 2012, 33, 1848.e15-1848.e20.	1.5	76
31	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.	1.8	76
32	Immunosuppressive treatment in refractory chronic inflammatory demyelinating polyradiculoneuropathy. A nationwide retrospective analysis. European Journal of Neurology, 2011, 18, 1417-1421.	1.7	71
33	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.	0.9	70
34	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.	1.0	64
35	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.	0.3	60
36	Differential electrophysiological features of neuropathies associated with 17p11.2 deletion and duplication. Muscle and Nerve, 1995, 18, 628-635.	1.0	60

#	Article	IF	Citations
37	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.	1.3	57
38	Botulinum toxin B ultrasound-guided injections for sialorrhea in amyotrophic lateral sclerosis and Parkinson's disease. Parkinsonism and Related Disorders, 2007, 13, 299-303.	1.1	55
39	Ultrasound visualization of nerve morphological alteration at the site of conduction block. Muscle and Nerve, 2009, 40, 1068-1070.	1.0	54
40	ATXN2 polyQ intermediate repeats are a modifier of ALS survival. Neurology, 2015, 84, 251-258.	1.5	52
41	Neuromyopathy and restrictive cardiomyopathy with accumulation of intermediate filaments: a clinical, morphological and biochemical study. Acta Neuropathologica, 1991, 81, 632-640.	3.9	51
42	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.	1.4	51
43	â€˜Behr syndromeâ€™ with OPA1 compound heterozygote mutations. Brain, 2015, 138, e321-e321.	3.7	50
44	Nutritional and metabolic support in patients with amyotrophic lateral sclerosis. Nutrition, 2012, 28, 959-966.	1.1	48
45	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.	1.6	48
46	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.	1.0	46
47	Hypermyelinating neuropathy, mental retardation and epilepsy in a case of merosin deficiency. Neuromuscular Disorders, 2002, 12, 392-398.	0.3	46
48	Elevated Levels of Selenium Species in Cerebrospinal Fluid of Amyotrophic Lateral Sclerosis Patients with Disease-Associated Gene Mutations. Neurodegenerative Diseases, 2017, 17, 171-180.	0.8	46
49	An Italian family with inclusionâ€body myopathy and frontotemporal dementia due to mutation in the VCP gene. Muscle and Nerve, 2008, 37, 111-114.	1.0	44
50	CHCH10 mutations in an Italian cohort of familial and sporadic amyotrophic lateral sclerosis patients. Neurobiology of Aging, 2015, 36, 1767.e3-1767.e6.	1.5	44
51	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.	0.8	44
52	Repetitive transcranial magnetic stimulation for ALS. Neuroscience Letters, 2006, 408, 135-140.	1.0	43
53	TTR-related amyloid neuropathy: clinical, electrophysiological and pathological findings in 15 unrelated patients. Neurological Sciences, 2013, 34, 1057-1063.	0.9	43
54	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

#	Article	IF	Citations
55	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.	1.4	42
56	A novel HSPB1 mutation in an Italian patient with CMT2/dHMN phenotype. Journal of the Neurological Sciences, 2010, 298, 114-117.	0.3	42
57	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.	1.5	42
58	Mitochondrial Network Genes in the Skeletal Muscle of Amyotrophic Lateral Sclerosis Patients. PLoS ONE, 2013, 8, e57739.	1.1	42
59	TBK1 is associated with ALS and ALS-FTD in Sardinian patients. Neurobiology of Aging, 2016, 43, 180.e1-180.e5.	1.5	40
60	Motor cortex stimulation for amyotrophic lateral sclerosis. Time for a therapeutic trial?. Clinical Neurophysiology, 2004, 115, 1479-1485.	0.7	38
61	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.	1.5	38
62	Chronic progressive steroid responsive axonal polyneuropathy: A CIDP variant or a primary axonal disorder?. , 1996, 19, 365-371.		37
63	Interferon-alpha may benefit steroid unresponsive chronic inflammatory demyelinating polyneuropathy.. Journal of Neurology, Neurosurgery and Psychiatry, 1995, 58, 638-639.	0.9	36
64	Consistent bone marrow-derived cell mobilization following repeated short courses of granulocyteâ€“colony-stimulating factor in patients with amyotrophic lateral sclerosis: results from a multicenter prospective trial. Cytotherapy, 2010, 12, 50-59.	0.3	36
65	A longitudinal study defined circulating microRNAs as reliable biomarkers for disease prognosis and progression in ALS human patients. Cell Death Discovery, 2021, 7, 4.	2.0	36
66	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, .	3.3	36
67	Matrin 3 variants are frequent in Italian ALS patients. Neurobiology of Aging, 2017, 49, 218.e1-218.e7.	1.5	35
68	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.	1.4	35
69	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.	2.8	35
70	Peripheral neuropathy with hypomyelinating features in adult-onset Krabbe's disease. Neuromuscular Disorders, 2002, 12, 386-391.	0.3	34
71	Nerve ultrasound findings in neuropathy associated with antiâ€myelinâ€associated glycoprotein antibodies. European Journal of Neurology, 2015, 22, 193-202.	1.7	34
72	ATXN1 intermediate-length polyglutamine expansions are associated with amyotrophic lateral sclerosis. Neurobiology of Aging, 2018, 64, 157.e1-157.e5.	1.5	34

#	Article	IF	Citations
73	Motor cortex stimulation for ALS: A double blind placebo-controlled study. Neuroscience Letters, 2009, 464, 18-21.	1.0	33
74	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.	1.1	33
75	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.	1.5	32
76	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.	2.3	32
77	Ultrasound evaluation in transthyretinâ€related amyloid neuropathy. Muscle and Nerve, 2014, 50, 372-376.	1.0	32
78	Clinical, neurophysiological and pathological findings of HNPP patients with 17p12 deletion: A single-centre experience. Journal of the Neurological Sciences, 2014, 341, 46-50.	0.3	32
79	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.	1.4	31
80	Early diagnosis followed by front-line autologous peripheral blood stem cell transplantation for patients affected by POEMS syndrome. Leukemia Research, 2008, 32, 1309-1312.	0.4	30
81	Primary multifocal lymphoma of peripheral nervous system: Case report and review of the literature. Muscle and Nerve, 2014, 50, 1016-1022.	1.0	30
82	Flow Cytofluorimetric Analysis of Anti-LRP4 (LDL Receptor-Related Protein 4) Autoantibodies in Italian Patients with Myasthenia Gravis. PLoS ONE, 2015, 10, e0135378.	1.1	30
83	Hereditary motor and sensory neuropathy with calf hypertrophy is associated with 17p 11.2 duplication. Annals of Neurology, 1994, 35, 552-558.	2.8	29
84	Neuromyelitis optica spectrum disorder as a paraneoplastic manifestation of lung adenocarcinoma expressing aquaporin-4. Multiple Sclerosis Journal, 2015, 21, 791-794.	1.4	28
85	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.	1.4	28
86	New ALSâ€Related Genes Expand the Spectrum Paradigm of Amyotrophic Lateral Sclerosis. Brain Pathology, 2016, 26, 266-275.	2.1	26
87	Efficacy of lenalidomide plus dexamethasone for POEMS syndrome relapsed after autologous peripheral stemâ€cell transplantation. American Journal of Hematology, 2012, 87, 641-642.	2.0	24
88	The S100A4 Transcriptional Inhibitor Niclosamide Reduces Pro-Inflammatory and Migratory Phenotypes of Microglia: Implications for Amyotrophic Lateral Sclerosis. Cells, 2019, 8, 1261.	1.8	24
89	hATTR Pathology: Nerve Biopsy Results from Italian Referral Centers. Brain Sciences, 2020, 10, 780.	1.1	24
90	Light chain deposition in peripheral nerve as a cause of mononeuritis multiplex in WaldenstrÃ¶m's macroglobulinaemia. Journal of the Neurological Sciences, 2010, 291, 89-91.	0.3	23

#	Article	IF	Citations
91	Non-coding variants contribute to the clinical heterogeneity of TTR amyloidosis. European Journal of Human Genetics, 2017, 25, 1055-1060.	1.4	23
92	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.	0.9	22
93	Heterozygous SOD1 D90A mutation presenting as slowly progressive predominant upper motor neuron amyotrophic lateral sclerosis. Neurological Sciences, 2009, 30, 517-520.	0.9	21
94	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.	1.5	21
95	Giant axonal neuropathy: report on a case with focal fiber loss. Acta Neuropathologica, 1992, 83, 543-546.	3.9	20
96	Long-term motor cortex stimulation for amyotrophic lateral sclerosis. Brain Stimulation, 2010, 3, 22-27.	0.7	20
97	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.	1.4	20
98	Comorbidity of dementia with amyotrophic lateral sclerosis (ALS): insights from a large multicenter Italian cohort. Journal of Neurology, 2017, 264, 2224-2231.	1.8	19
99	Histamine beyond its effects on allergy: Potential therapeutic benefits for the treatment of Amyotrophic Lateral Sclerosis (ALS). , 2019, 202, 120-131.		19
100	Clinical and pathological heterogeneity in a series of 31 patients with IgM-related neuropathy. Journal of the Neurological Sciences, 2012, 319, 75-80.	0.3	18
101	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.	0.6	18
102	ALS skin fibroblasts reveal oxidative stress and ERK1/2-mediated cytoplasmic localization of TDP-43. Cellular Signalling, 2020, 70, 109591.	1.7	18
103	Renal Involvement in Hereditary Transthyretin Amyloidosis: An Italian Single-Centre Experience. Brain Sciences, 2021, 11, 980.	1.1	18
104	Autosomal recessive hypermyelinating neuropathy. Acta Neuropathologica, 1994, 87, 337-342.	3.9	17
105	Sural nerve pathology in ALS patients: a single-centre experience. Neurological Sciences, 2012, 33, 1095-1099.	0.9	17
106	Clinicalâ€“neurophysiological correlations in a series of patients with IgM-related neuropathy. Clinical Neurophysiology, 2013, 124, 1899-1903.	0.7	17
107	Start codon mutation of GYG1 causing late-onset polyglucosan body myopathy with nemaline rods. Journal of Neurology, 2016, 263, 2133-2135.	1.8	17
108	SEIPIN S90L Mutation in an Italian family with CMT2/dHMN and pyramidal signs. Muscle and Nerve, 2010, 42, 448-451.	1.0	16

#	Article	IF	Citations
109	A novel homozygous mutation in the <scp>MTMR2</scp> gene in two siblings with â€˜hypermyelinating neuropathyâ€™. Journal of the Peripheral Nervous System, 2013, 18, 192-194.	1.4	16
110	A useful electrophysiological test for diagnosis of minimal conduction block. Clinical Neurophysiology, 2001, 112, 1041-1048.	0.7	15
111	Occurrence of nerve entrapment lesion in chronic inflammatory demyelinating polyneuropathy. Clinical Neurophysiology, 2004, 115, 1140-1144.	0.7	15
112	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.	0.3	15
113	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.	0.3	15
114	Influence of comorbidities on the phenotype of patients affected by Charcotâ€“Marieâ€“Tooth neuropathy type 1A. Neuromuscular Disorders, 2013, 23, 902-906.	0.3	15
115	Admission neurophysiological abnormalities in Guillainâ€“BarrÃ© syndrome: A single-center experience. Clinical Neurology and Neurosurgery, 2015, 135, 6-10.	0.6	15
116	High-Throughput Genetic Testing in ALS: The Challenging Path of Variant Classification Considering the ACMG Guidelines. Genes, 2020, 11, 1123.	1.0	15
117	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.	1.4	15
118	Chronic inflammatory demyelinating polyradiculoneuropathy: can a diagnosis be made in patients not fulfilling electrodiagnostic criteria?. European Journal of Neurology, 2021, 28, 620-629.	1.7	15
119	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.	0.5	14
120	A new singleâ€nucleotide deletion of PMP22 in an HNPP family without recurrent palsies. Muscle and Nerve, 2008, 38, 1060-1064.	1.0	14
121	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.	0.7	14
122	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.	0.9	14
123	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.	0.6	13
124	Teaching NeuroImages: Peroneal intraneural ganglion cyst: A rare cause of drop foot in a child. Neurology, 2012, 78, e46-e47.	1.5	13
125	Frontotemporal dementia, Parkinsonism and lower motor neuron involvement in a patient with C9ORF72 expansion. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 66-69.	1.1	13
126	Epigenetic profiling of Italian patients identified methylation sites associated with hereditary transthyretin amyloidosis. Clinical Epigenetics, 2020, 12, 176.	1.8	13

#	Article	IF	Citations
127	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.	1.7	13
128	SOD1 G93D mutation presenting as paucisymptomatic amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2009, 10, 479-482.	2.3	12
129	D11Y SOD1 mutation and benign ALS: A consistent genotype-phenotype correlation. Journal of the Neurological Sciences, 2011, 309, 31-33.	0.3	12
130	A novel compound heterozygousALS2mutation in two Italian siblings with juvenile amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 470-472.	1.1	12
131	Sural nerve biopsy in peripheral neuropathies: 30-year experience from a single center. Neurological Sciences, 2020, 41, 341-346.	0.9	12
132	Restless Leg Syndrome in Different Types of Demyelinating Neuropathies: A Single-Center Pilot Study. Journal of Clinical Sleep Medicine, 2013, 09, 945-949.	1.4	12
133	A novel L67P SOD1 mutation in an Italian ALS patient. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2011, 12, 150-152.	2.3	11
134	Letter: faecal microbiota transplantation in combination with fidaxomicin to treat severe complicated recurrentClostridium difficileinfection. Alimentary Pharmacology and Therapeutics, 2015, 42, 1030-1030.	1.9	11
135	Targeting S100A4 with niclosamide attenuates inflammatory and profibrotic pathways in models of amyotrophic lateral sclerosis. Journal of Neuroinflammation, 2021, 18, 132.	3.1	11
136	Neuropathy with Predominant Small Fiber Involvement Associated with Abnormal Anti-MAG Titer. Internal Medicine, 2010, 49, 2627-2629.	0.3	10
137	Pathological Findings in Chronic Inflammatory Demyelinating Polyradiculoneuropathy: A Single-Center Experience. Brain Sciences, 2020, 10, 383.	1.1	10
138	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.	0.9	10
139	Gastrointestinal Manifestations in Hereditary Transthyretin Amyloidosis: a Single-Centre Experience. Journal of Gastrointestinal and Liver Diseases, 2020, 29, 339-343.	0.5	10
140	Intravenous Immunoglobulin Treatment in Autoimmune Neurological Disordersâ€"Effects on Quality of Life. Human Immunology, 2005, 66, 417-421.	1.2	9
141	Progressive multifocal leukoencephalopathy in a patient with Franklin disease and hypogammaglobulinemia. Journal of the Neurological Sciences, 2009, 284, 203-204.	0.3	9
142	ATTRv in Lazio-Italy: A High-Prevalence Region in a Non-Endemic Country. Genes, 2021, 12, 829.	1.0	9
143	Teaching Neuro Images : The full-blown neuroimaging of Wernicke encephalopathy. Neurology, 2009, 72, e115.	1.5	8
144	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.	2.3	8

#	Article	IF	Citations
145	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.	0.3	8
146	Mutant human Î²4 subunit identified in amyotrophic lateral sclerosis patients impairs nicotinic receptor function. Pflugers Archiv European Journal of Physiology, 2011, 461, 225-233.	1.3	8
147	HFE p.H63D polymorphism does not influence ALS phenotype and survival. Neurobiology of Aging, 2015, 36, 2906.e7-2906.e11.	1.5	8
148	Patisiran in hATTR Amyloidosis: Six-Month Latency Period before Efficacy. Brain Sciences, 2021, 11, 515.	1.1	8
149	Ocular Involvement in Hereditary Transthyretin Amyloidosis: A Case Series Describing Novel Potential Biomarkers. Genes, 2021, 12, 927.	1.0	8
150	Exome Sequencing Reveals VCP Mutations as a Cause of Familial ALS. Neuron, 2011, 69, 397.	3.8	7
151	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.	2.3	7
152	MRI Neurography Findings in Patients with Idiopathic Brachial Plexopathy: Correlations with Clinical-neurophysiological Data in Eight Consecutive Cases. Internal Medicine, 2013, 52, 2031-2039.	0.3	7
153	Nerve ultrasound in patients with CMT1C: Description of 3 cases. Muscle and Nerve, 2015, 51, 781-782.	1.0	7
154	Novel variants and cellular studies on patientsâ€™ primary fibroblasts support a role for NEK1 missense variants in ALS pathogenesis. Human Molecular Genetics, 2021, 30, 65-71.	1.4	7
155	SLITRK2, an X-linked modifier of the age at onset in C9orf72 frontotemporal lobar degeneration. Brain, 2021, 144, 2798-2811.	3.7	7
156	Acute axonal idiopathic polyneuropathy: A Guillain-BarrÃ© syndrome variant?. Italian Journal of Neurological Sciences, 1992, 13, 481-486.	0.1	6
157	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.	0.9	6
158	Clinical, electrophysiological and pathological findings in a patient with Charcotâ€“Marieâ€“Tooth disease 4D caused by the NDRG1 Lom mutation. Journal of the Neurological Sciences, 2014, 345, 271-273.	0.3	6
159	Nerve ultrasound in CMT2E/CMT1F due to NEFL mutation: Confirmation of an axonal pathology. Clinical Neurophysiology, 2016, 127, 2990-2991.	0.7	6
160	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.	1.5	6
161	Monophasic demyelinating disease of the central nervous system associated with Hepatitis A infection. Journal of Neurology, 2006, 253, 944-945.	1.8	5
162	Recurrent miller fisher: a new case report and a literature review. Clinica Terapeutica, 2017, 168, e208-e213.	0.2	5

#	Article	IF	Citations
163	NEUROLOGIC IMPROVEMENT AFTER PERIPHERAL BLOOD STEM CELL TRANSPLANTATION IN POEMS. Neurology, 2009, 73, 1165-1166.	1.5	4
164	Posterior ischaemic myelopathy associated with cocaine abuse. Internal Medicine Journal, 2010, 40, 732-733.	0.5	4
165	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.	2.3	4
166	Tuberculous nephritis accompanying neuromyelitis optica: causal or coincidental association?. Journal of Neurology, 2014, 261, 1028-1030.	1.8	4
167	SOD1 p.D12Y variant is associated with amyotrophic lateral sclerosis/distal myopathy spectrum. European Journal of Neurology, 2020, 27, 1304-1309.	1.7	4
168	Genome-Wide Analyses Identify KIF5A as a Novel ALS Gene. SSRN Electronic Journal, 0, , .	0.4	4
169	Teaching Neuro Image : MRI of diabetic lumbar plexopathy treated with local steroid injection. Neurology, 2009, 72, e32-3.	1.5	3
170	TEACHING NEURO IMAGES : THE FULL-BLOWN NEUROIMAGING OF WERNICKE ENCEPHALOPATHY. Neurology, 2010, 74, 527-528.	1.5	3
171	Progressive ascending myelopathy: atypical forms of multiple sclerosis or what else?. Journal of Neurology, 2011, 258, 1965-1970.	1.8	3
172	Generation and characterization of a human iPSC line from an ALS patient carrying the Q66K-MATR3 mutation. Stem Cell Research, 2018, 33, 146-150.	0.3	3
173	Personalized Prevention in Mercury-Induced Amyotrophic Lateral Sclerosis: A Case Report. Applied Sciences (Switzerland), 2020, 10, 7839.	1.3	3
174	A compound score to screen patients with hereditary transthyretin amyloidosis. Journal of Neurology, 2022, , .	1.8	3
175	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.	2.2	3
176	MOG autoimmunity mimicking CLIPPERS syndrome: Case report and literature review. Journal of Neuroimmunology, 2022, 367, 577875.	1.1	3
177	Demyelinating encephalomyeloradiculitis with BalÃ²-like lesions. Journal of Neurology, 2010, 257, 1566-1567.	1.8	2
178	A novel GJB1 mutation in an Italian patient with Charcotâ€“Marieâ€“Tooth disease and pyramidal signs. Muscle and Nerve, 2011, 44, 613-615.	1.0	2
179	Peripheral neuropathy and 46XY gonadal dysgenesis: Confirmation of a heterogeneous entity. Clinical Neurology and Neurosurgery, 2012, 114, 748-750.	0.6	2
180	Cranial botulism. Neuromuscular Disorders, 2012, 22, 995-996.	0.3	2

#	Article	IF	Citations
181	Distinct lymphocytes subsets in IgM-related neuropathy: clinical-immunological correlations. Neurological Sciences, 2015, 36, 303-308.	0.9	2
182	‘White Nails'. European Neurology, 2015, 73, 89-89.	0.6	2
183	Small Fibre Involvement in Multifocal Motor Neuropathy Explored with Sudoscan: A Single-Centre Experience. Diagnostics, 2020, 10, 755.	1.3	2
184	Abnormal vascular smooth muscle cell proliferation in sural nerve biopsy from a patient with sensorimotor axonal neuropathy. Neuropathology, 2011, 31, 197-198.	0.7	1
185	Teaching Neuro Images : Cochleitis. Neurology, 2011, 77, e109.	1.5	1
186	PERIPHERAL NERVOUS SYSTEM INVOLVEMENT IN LYMPHOPROLIFERATIVE DISORDERS. Mediterranean Journal of Hematology and Infectious Diseases, 2018, 10, e2018057.	0.5	1
187	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.	1.4	1
188	Response to: SOD1 mutations in adultâ€onset distal spinal muscular atrophy. European Journal of Neurology, 2020, 27, e74.	1.7	1
189	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.	0.3	1
190	Prolonged distal motor latency of median nerve does not improve diagnostic accuracy for CIDP. Journal of Neurology, 2021, , 1.	1.8	1
191	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.	0.3	1
192	Reply to Dr van Dijk. Clinical Neurophysiology, 2001, 112, 2164-2165.	0.7	0
193	A Useful Electrophysiologic Test For Diagnosis Of Minimal Conduction Block. Journal of the Peripheral Nervous System, 2001, 6, 54-55.	1.4	0
194	Multinevritis of cranial nerves following inhalation of toxins. Neurological Research, 2003, 25, 208-210.	0.6	0
195	Is carpal tunnel syndrome surgery useful in patients with diabetes or autoimmune polyneuropathies?. Journal of the Peripheral Nervous System, 2004, 9, 109-109.	1.4	0
196	NATURAL HISTORY OF YOUNG-ADULT AMYOTROPHIC LATERAL SCLEROSIS. Neurology, 2009, 73, 648-650.	1.5	0
197	Retinal Detachment with an Unusual Shape. Internal Medicine, 2009, 48, 1777-1778.	0.3	0
198	Skin Changes in POEMS Syndrome. European Neurology, 2015, 73, 112-112.	0.6	0

#	Article	IF	Citations
199	Thr124Met myelin protein zero mutation mimicking motor neuron disease. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2021, , 1-6.	1.1	0
200	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.	0.3	0
201	Guillainâ€“BarrÃ© syndrome from an emergency department view: how to better predict the outcome?. Neurological Research, 2022, , 1-5.	0.6	0