

Mario Sabatelli

List of Publications by Year in descending order

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Version: 2024-02-01

201
papers

13,103
citations

53794

45
h-index

25787

108
g-index

205
all docs

205
docs citations

205
times ranked

12558
citing authors

#	ARTICLE	IF	CITATIONS
1	A Hexanucleotide Repeat Expansion in C9ORF72 Is the Cause of Chromosome 9p21-Linked ALS-FTD. <i>Neuron</i> , 2011, 72, 257-268.	8.1	3,833
2	Exome Sequencing Reveals VCP Mutations as a Cause of Familial ALS. <i>Neuron</i> , 2010, 68, 857-864.	8.1	1,100
3	Frequency of the C9orf72 hexanucleotide repeat expansion in patients with amyotrophic lateral sclerosis and frontotemporal dementia: a cross-sectional study. <i>Lancet Neurology</i> , The, 2012, 11, 323-330.	10.2	1,039
4	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. <i>Neuron</i> , 2018, 97, 1268-1283.e6.	8.1	517
5	Mutations in the Matrin 3 gene cause familial amyotrophic lateral sclerosis. <i>Nature Neuroscience</i> , 2014, 17, 664-666.	14.8	398
6	Intravenous immunoglobulin versus intravenous methylprednisolone for chronic inflammatory demyelinating polyradiculoneuropathy: a randomised controlled trial. <i>Lancet Neurology</i> , The, 2012, 11, 493-502.	10.2	185
7	Clinical characteristics of patients with familial amyotrophic lateral sclerosis carrying the pathogenic GGGGCC hexanucleotide repeat expansion of C9ORF72. <i>Brain</i> , 2012, 135, 784-793.	7.6	182
8	Heterogeneity of root and nerve ultrasound pattern in CIDP patients. <i>Clinical Neurophysiology</i> , 2014, 125, 160-165.	1.5	142
9	A Genome-Wide Association Study of Myasthenia Gravis. <i>JAMA Neurology</i> , 2015, 72, 396.	9.0	139
10	Two Italian kindreds with familial amyotrophic lateral sclerosis due to FUS mutation. <i>Neurobiology of Aging</i> , 2009, 30, 1272-1275.	3.1	128
11	P525L FUS mutation is consistently associated with a severe form of juvenile Amyotrophic Lateral Sclerosis. <i>Neuromuscular Disorders</i> , 2012, 22, 73-75.	0.6	124
12	Autosomal recessive hereditary motor and sensory neuropathy with focally folded myelin sheaths. <i>Neurology</i> , 1996, 46, 1318-1318.	1.1	121
13	Antibodies to neurofascin, contactin-1, and contactin-associated protein 1 in CIDP. <i>Neurology: Neuroimmunology and NeuroInflammation</i> , 2020, 7, .	6.0	118
14	A nationwide retrospective analysis on the effect of immune therapies in patients with chronic inflammatory demyelinating polyradiculoneuropathy. <i>European Journal of Neurology</i> , 2010, 17, 289-294.	3.3	115
15	Mutations in the CHCHD10 gene are a common cause of familial amyotrophic lateral sclerosis. <i>Brain</i> , 2014, 137, e311-e311.	7.6	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. <i>Movement Disorders</i> , 2011, 26, 313-319.	3.9	111
17	A two-stage genome-wide association study of sporadic amyotrophic lateral sclerosis. <i>Human Molecular Genetics</i> , 2009, 18, 1524-1532.	2.9	106
18	Rituximab in patients with chronic inflammatory demyelinating polyradiculoneuropathy: a report of 13 cases and review of the literature. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2011, 82, 306-308.	1.9	106

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19	Pure motor chronic inflammatory demyelinating polyneuropathy. <i>Journal of Neurology</i> , 2001, 248, 772-777.	3.6	102
20	An ALS-associated mutation in the FUS 3' UTR disrupts a microRNA-FUS regulatory circuitry. <i>Nature Communications</i> , 2014, 5, 4335.	12.8	102
21	Contribution of major amyotrophic lateral sclerosis genes to the etiology of sporadic disease. <i>Neurology</i> , 2012, 79, 66-72.	1.1	99
22	Genetic counselling in ALS: facts, uncertainties and clinical suggestions. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2014, 85, 478-485.	1.9	99
23	Factors predicting survival in ALS: a multicenter Italian study. <i>Journal of Neurology</i> , 2017, 264, 54-63.	3.6	96
24	Mutations in the 5' untranslated region of FUS causing FUS overexpression are associated with amyotrophic lateral sclerosis. <i>Human Molecular Genetics</i> , 2013, 22, 4748-4755.	2.9	94
25	Clinical and genetic heterogeneity of amyotrophic lateral sclerosis. <i>Clinical Genetics</i> , 2013, 83, 408-416.	2.0	92
26	Lithium carbonate in amyotrophic lateral sclerosis. <i>Neurology</i> , 2010, 75, 619-625.	1.1	90
27	Natural history of young-adult amyotrophic lateral sclerosis. <i>Neurology</i> , 2008, 71, 876-881.	1.1	81
28	FUS mutations in sporadic amyotrophic lateral sclerosis. <i>Neurobiology of Aging</i> , 2011, 32, 550.e1-550.e4.	3.1	79
29	<p>Diagnosis and Treatment of Hereditary Transthyretin Amyloidosis (hATTR) Polyneuropathy: Current Perspectives on Improving Patient Care<p>. <i>Therapeutics and Clinical Risk Management</i> , 2020, Volume 16, 109-123.	2.0	78
30	C9ORF72 hexanucleotide repeat expansions in the Italian sporadic ALS population. <i>Neurobiology of Aging</i> , 2012, 33, 1848.e15-1848.e20.	3.1	76
31	Monitoring effectiveness and safety of Tafamidis in transthyretin amyloidosis in Italy: a longitudinal multicenter study in a non-endemic area. <i>Journal of Neurology</i> , 2016, 263, 916-924.	3.6	76
32	Immunosuppressive treatment in refractory chronic inflammatory demyelinating polyradiculoneuropathy. A nationwide retrospective analysis. <i>European Journal of Neurology</i> , 2011, 18, 1417-1421.	3.3	71
33	Frequency and time to relapse after discontinuing 6-month therapy with IVlg or pulsed methylprednisolone in CIDP. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2015, 86, 729-734.	1.9	70
34	Repeated courses of granulocyte colony-stimulating factor in amyotrophic lateral sclerosis: Clinical and biological results from a prospective multicenter study. <i>Muscle and Nerve</i> , 2011, 43, 189-195.	2.2	64
35	Peripheral neuropathy with giant axons and cardiomyopathy associated with desmin type intermediate filaments in skeletal muscle. <i>Journal of the Neurological Sciences</i> , 1992, 109, 1-10.	0.6	60
36	Differential electrophysiological features of neuropathies associated with 17p11.2 deletion and duplication. <i>Muscle and Nerve</i> , 1995, 18, 628-635.	2.2	60

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37	Peripheral sensory-motor polyneuropathy, pigmentary retinopathy, and fatal cardiomyopathy in long-chain 3-hydroxy-acyl-CoA dehydrogenase deficiency. <i>European Journal of Pediatrics</i> , 1992, 151, 121-126.	2.7	57
38	Botulinum toxin B ultrasound-guided injections for sialorrhea in amyotrophic lateral sclerosis and Parkinson's disease. <i>Parkinsonism and Related Disorders</i> , 2007, 13, 299-303.	2.2	55
39	Ultrasound visualization of nerve morphological alteration at the site of conduction block. <i>Muscle and Nerve</i> , 2009, 40, 1068-1070.	2.2	54
40	<i>ATXN2</i> polyQ intermediate repeats are a modifier of ALS survival. <i>Neurology</i> , 2015, 84, 251-258.	1.1	52
41	Neuromyopathy and restrictive cardiomyopathy with accumulation of intermediate filaments: a clinical, morphological and biochemical study. <i>Acta Neuropathologica</i> , 1991, 81, 632-640.	7.7	51
42	ATTRv amyloidosis Italian Registry: clinical and epidemiological data. <i>Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis</i> , 2020, 27, 259-265.	3.0	51
43	â€ˆBehr syndromeâ€™™ with OPA1 compound heterozygote mutations. <i>Brain</i> , 2015, 138, e321-e321.	7.6	50
44	Nutritional and metabolic support in patients with amyotrophic lateral sclerosis. <i>Nutrition</i> , 2012, 28, 959-966.	2.4	48
45	Potential therapeutic targets for ALS: MIR206, MIR208b and MIR499 are modulated during disease progression in the skeletal muscle of patients. <i>Scientific Reports</i> , 2017, 7, 9538.	3.3	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. <i>Muscle and Nerve</i> , 1994, 17, 667-674.	2.2	46
47	Hypermyelinating neuropathy, mental retardation and epilepsy in a case of merosin deficiency. <i>Neuromuscular Disorders</i> , 2002, 12, 392-398.	0.6	46
48	Elevated Levels of Selenium Species in Cerebrospinal Fluid of Amyotrophic Lateral Sclerosis Patients with Disease-Associated Gene Mutations. <i>Neurodegenerative Diseases</i> , 2017, 17, 171-180.	1.4	46
49	An Italian family with inclusionâ€™body myopathy and frontotemporal dementia due to mutation in the <i>VCP</i> gene. <i>Muscle and Nerve</i> , 2008, 37, 111-114.	2.2	44
50	CHCH10 mutations in an Italian cohort of familial and sporadic amyotrophic lateral sclerosis patients. <i>Neurobiology of Aging</i> , 2015, 36, 1767.e3-1767.e6.	3.1	44
51	Proteostasis and ALS: protocol for a phase II, randomised, double-blind, placebo-controlled, multicentre clinical trial for colchicine in ALS (Co-ALS). <i>BMJ Open</i> , 2019, 9, e028486.	1.9	44
52	Repetitive transcranial magnetic stimulation for ALS. <i>Neuroscience Letters</i> , 2006, 408, 135-140.	2.1	43
53	TTR-related amyloid neuropathy: clinical, electrophysiological and pathological findings in 15 unrelated patients. <i>Neurological Sciences</i> , 2013, 34, 1057-1063.	1.9	43
54	Sensitivity and specificity of diagnostic criteria for conduction block in chronic inflammatory demyelinating polyneuropathy. <i>Electroencephalography and Clinical Neurophysiology - Evoked Potentials</i> , 1993, 89, 161-169.	2.0	42

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55	Rare missense variants of neuronal nicotinic acetylcholine receptor altering receptor function are associated with sporadic amyotrophic lateral sclerosis. <i>Human Molecular Genetics</i> , 2009, 18, 3997-4006.	2.9	42
56	A novel HSPB1 mutation in an Italian patient with CMT2/dHMN phenotype. <i>Journal of the Neurological Sciences</i> , 2010, 298, 114-117.	0.6	42
57	Primary fibroblasts cultures reveal TDP-43 abnormalities in amyotrophic lateral sclerosis patients with and without SOD1 mutations. <i>Neurobiology of Aging</i> , 2015, 36, 2005.e5-2005.e13.	3.1	42
58	Mitochondrial Network Genes in the Skeletal Muscle of Amyotrophic Lateral Sclerosis Patients. <i>PLoS ONE</i> , 2013, 8, e57739.	2.5	42
59	TBK1 is associated with ALS and ALS-FTD in Sardinian patients. <i>Neurobiology of Aging</i> , 2016, 43, 180.e1-180.e5.	3.1	40
60	Motor cortex stimulation for amyotrophic lateral sclerosis. Time for a therapeutic trial?. <i>Clinical Neurophysiology</i> , 2004, 115, 1479-1485.	1.5	38
61	Relations between C9orf72 expansion size in blood, age at onset, age at collection and transmission across generations in patients and presymptomatic carriers. <i>Neurobiology of Aging</i> , 2019, 74, 234.e1-234.e8.	3.1	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.. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 1995, 58, 638-639.	1.9	36
64	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. <i>Cytotherapy</i> , 2010, 12, 50-59.	0.7	36
65	A longitudinal study defined circulating microRNAs as reliable biomarkers for disease prognosis and progression in ALS human patients. <i>Cell Death Discovery</i> , 2021, 7, 4.	4.7	36
66	Identification of genetic risk loci and prioritization of genes and pathways for myasthenia gravis: a genome-wide association study. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2022, 119, .	7.1	36
67	Matrin 3 variants are frequent in Italian ALS patients. <i>Neurobiology of Aging</i> , 2017, 49, 218.e1-218.e7.	3.1	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. <i>Frontiers in Neuroscience</i> , 2019, 13, 485.	2.8	35
69	A multicenter retrospective study of charcot-Marie-Tooth disease type 4B (CMT4B) associated with mutations in myotubularin-related proteins (MTMRs). <i>Annals of Neurology</i> , 2019, 86, 55-67.	5.3	35
70	Peripheral neuropathy with hypomyelinating features in adult-onset Krabbe's disease. <i>Neuromuscular Disorders</i> , 2002, 12, 386-391.	0.6	34
71	Nerve ultrasound findings in neuropathy associated with anti-myelin-associated glycoprotein antibodies. <i>European Journal of Neurology</i> , 2015, 22, 193-202.	3.3	34
72	ATXN1 intermediate-length polyglutamine expansions are associated with amyotrophic lateral sclerosis. <i>Neurobiology of Aging</i> , 2018, 64, 157.e1-157.e5.	3.1	34

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73	Motor cortex stimulation for ALS: A double blind placebo-controlled study. <i>Neuroscience Letters</i> , 2009, 464, 18-21.	2.1	33
74	Epstein-Barr virus antibodies in serum and cerebrospinal fluid from Multiple sclerosis, Chronic Inflammatory Demyelinating Polyradiculoneuropathy and Amyotrophic Lateral Sclerosis. <i>Journal of Neuroimmunology</i> , 2010, 225, 149-152.	2.3	33
75	SOD1 G93D sporadic amyotrophic lateral sclerosis (SALS) patient with rapid progression and concomitant novel ANG variant. <i>Neurobiology of Aging</i> , 2011, 32, 1924.e15-1924.e18.	3.1	32
76	Uncovering amyotrophic lateral sclerosis phenotypes: Clinical features and long-term follow-up of upper motor neuron-dominant ALS. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2011, 12, 278-282.	2.1	32
77	Ultrasound evaluation in transthyretin-related amyloid neuropathy. <i>Muscle and Nerve</i> , 2014, 50, 372-376.	2.2	32
78	Clinical, neurophysiological and pathological findings of HNPP patients with 17p12 deletion: A single-centre experience. <i>Journal of the Neurological Sciences</i> , 2014, 341, 46-50.	0.6	32
79	pSTAT1, pSTAT3, and Tbet as markers of disease activity in chronic inflammatory demyelinating polyradiculoneuropathy. <i>Journal of the Peripheral Nervous System</i> , 2009, 14, 107-117.	3.1	31
80	Early diagnosis followed by front-line autologous peripheral blood stem cell transplantation for patients affected by POEMS syndrome. <i>Leukemia Research</i> , 2008, 32, 1309-1312.	0.8	30
81	Primary multifocal lymphoma of peripheral nervous system: Case report and review of the literature. <i>Muscle and Nerve</i> , 2014, 50, 1016-1022.	2.2	30
82	Flow Cytofluorimetric Analysis of Anti-LRP4 (LDL Receptor-Related Protein 4) Autoantibodies in Italian Patients with Myasthenia Gravis. <i>PLoS ONE</i> , 2015, 10, e0135378.	2.5	30
83	Hereditary motor and sensory neuropathy with calf hypertrophy is associated with 17p 11.2 duplication. <i>Annals of Neurology</i> , 1994, 35, 552-558.	5.3	29
84	Neuromyelitis optica spectrum disorder as a paraneoplastic manifestation of lung adenocarcinoma expressing aquaporin-4. <i>Multiple Sclerosis Journal</i> , 2015, 21, 791-794.	3.0	28
85	Sudoscan in the evaluation and follow-up of patients and carriers with TTR mutations: experience from an Italian Centre. <i>Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis</i> , 2018, 25, 242-246.	3.0	28
86	New ALS-Related Genes Expand the Spectrum Paradigm of Amyotrophic Lateral Sclerosis. <i>Brain Pathology</i> , 2016, 26, 266-275.	4.1	26
87	Efficacy of lenalidomide plus dexamethasone for POEMS syndrome relapsed after autologous peripheral stem cell transplantation. <i>American Journal of Hematology</i> , 2012, 87, 641-642.	4.1	24
88	The S100A4 Transcriptional Inhibitor Niclosamide Reduces Pro-Inflammatory and Migratory Phenotypes of Microglia: Implications for Amyotrophic Lateral Sclerosis. <i>Cells</i> , 2019, 8, 1261.	4.1	24
89	hATTR Pathology: Nerve Biopsy Results from Italian Referral Centers. <i>Brain Sciences</i> , 2020, 10, 780.	2.3	24
90	Light chain deposition in peripheral nerve as a cause of mononeuritis multiplex in Waldenström's macroglobulinaemia. <i>Journal of the Neurological Sciences</i> , 2010, 291, 89-91.	0.6	23

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91	Non-coding variants contribute to the clinical heterogeneity of TTR amyloidosis. <i>European Journal of Human Genetics</i> , 2017, 25, 1055-1060.	2.8	23
92	Classification of familial amyotrophic lateral sclerosis by family history: effects on frequency of genes mutation. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2012, 83, 1201-1203.	1.9	22
93	Heterozygous SOD1 D90A mutation presenting as slowly progressive predominant upper motor neuron amyotrophic lateral sclerosis. <i>Neurological Sciences</i> , 2009, 30, 517-520.	1.9	21
94	Coexistence of variants in TBK1 and in other ALS-related genes elucidates an oligogenic model of pathogenesis in sporadic ALS. <i>Neurobiology of Aging</i> , 2019, 84, 239.e9-239.e14.	3.1	21
95	Giant axonal neuropathy: report on a case with focal fiber loss. <i>Acta Neuropathologica</i> , 1992, 83, 543-546.	7.7	20
96	Long-term motor cortex stimulation for amyotrophic lateral sclerosis. <i>Brain Stimulation</i> , 2010, 3, 22-27.	1.6	20
97	AL amyloid neuropathy mimicking a chronic inflammatory demyelinating polyneuropathy. <i>Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis</i> , 2012, 19, 53-55.	3.0	20
98	Comorbidity of dementia with amyotrophic lateral sclerosis (ALS): insights from a large multicenter Italian cohort. <i>Journal of Neurology</i> , 2017, 264, 2224-2231.	3.6	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. <i>Journal of the Neurological Sciences</i> , 2012, 319, 75-80.	0.6	18
101	Charcot-Marie-Tooth type 2 and distal hereditary motor neuropathy: Clinical, neurophysiological and genetic findings from a single-centre experience. <i>Clinical Neurology and Neurosurgery</i> , 2016, 144, 67-71.	1.4	18
102	ALS skin fibroblasts reveal oxidative stress and ERK1/2-mediated cytoplasmic localization of TDP-43. <i>Cellular Signalling</i> , 2020, 70, 109591.	3.6	18
103	Renal Involvement in Hereditary Transthyretin Amyloidosis: An Italian Single-Centre Experience. <i>Brain Sciences</i> , 2021, 11, 980.	2.3	18
104	Autosomal recessive hypermyelinating neuropathy. <i>Acta Neuropathologica</i> , 1994, 87, 337-342.	7.7	17
105	Sural nerve pathology in ALS patients: a single-centre experience. <i>Neurological Sciences</i> , 2012, 33, 1095-1099.	1.9	17
106	Clinical neurophysiological correlations in a series of patients with IgM-related neuropathy. <i>Clinical Neurophysiology</i> , 2013, 124, 1899-1903.	1.5	17
107	Start codon mutation of GYG1 causing late-onset polyglucosan body myopathy with nemaline rods. <i>Journal of Neurology</i> , 2016, 263, 2133-2135.	3.6	17
108	<i>SEIPIN</i> S90L Mutation in an Italian family with CMT2/dHMN and pyramidal signs. <i>Muscle and Nerve</i> , 2010, 42, 448-451.	2.2	16

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109	A novel homozygous mutation in the <i>MTMR2</i> gene in two siblings with hypermyelinating neuropathy TM . <i>Journal of the Peripheral Nervous System</i> , 2013, 18, 192-194.	3.1	16
110	A useful electrophysiological test for diagnosis of minimal conduction block. <i>Clinical Neurophysiology</i> , 2001, 112, 1041-1048.	1.5	15
111	Occurrence of nerve entrapment lesion in chronic inflammatory demyelinating polyneuropathy. <i>Clinical Neurophysiology</i> , 2004, 115, 1140-1144.	1.5	15
112	Triple A syndrome: A novel compound heterozygous mutation in the AAAS gene in an Italian patient without adrenal insufficiency. <i>Journal of the Neurological Sciences</i> , 2010, 290, 150-152.	0.6	15
113	An Italian case of hereditary myopathy with early respiratory failure (HMERF) not associated with the titin kinase domain R279W mutation. <i>Neuromuscular Disorders</i> , 2010, 20, 730-734.	0.6	15
114	Influence of comorbidities on the phenotype of patients affected by Charcot-Marie-Tooth neuropathy type 1A. <i>Neuromuscular Disorders</i> , 2013, 23, 902-906.	0.6	15
115	Admission neurophysiological abnormalities in Guillain-Barré syndrome: A single-center experience. <i>Clinical Neurology and Neurosurgery</i> , 2015, 135, 6-10.	1.4	15
116	High-Throughput Genetic Testing in ALS: The Challenging Path of Variant Classification Considering the ACMG Guidelines. <i>Genes</i> , 2020, 11, 1123.	2.4	15
117	Relevance of diagnostic investigations in chronic inflammatory demyelinating polyradiculoneuropathy: Data from the Italian CIDP database. <i>Journal of the Peripheral Nervous System</i> , 2020, 25, 152-161.	3.1	15
118	Chronic inflammatory demyelinating polyradiculoneuropathy: can a diagnosis be made in patients not fulfilling electrodiagnostic criteria?. <i>European Journal of Neurology</i> , 2021, 28, 620-629.	3.3	15
119	Chronic GM2 gangliosidosis type Sandhoff associated with a novel missense HEXB gene mutation causing a double pathogenic effect. <i>Molecular Genetics and Metabolism</i> , 2007, 91, 111-114.	1.1	14
120	A new single nucleotide deletion of PMP22 in an HNPP family without recurrent palsies. <i>Muscle and Nerve</i> , 2008, 38, 1060-1064.	2.2	14
121	Usefulness of F-18 FDG PET/CT in the Follow-up of POEMS Syndrome After Autologous Peripheral Blood Stem Cell Transplantation. <i>Clinical Nuclear Medicine</i> , 2012, 37, 181-183.	1.3	14
122	Clinical features and outcomes of the flail arm and flail leg and pure lower motor neuron MND variants: a multicentre Italian study. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2020, 91, 1001-1003.	1.9	14
123	A case of CMT 1B due to Val 102/fs null mutation of the MPZ gene presenting as hyperCKemia. <i>Clinical Neurology and Neurosurgery</i> , 2010, 112, 794-797.	1.4	13
124	Teaching NeuroImages: Peroneal intraneural ganglion cyst: A rare cause of drop foot in a child. <i>Neurology</i> , 2012, 78, e46-e47.	1.1	13
125	Frontotemporal dementia, Parkinsonism and lower motor neuron involvement in a patient with C9ORF72 expansion. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2013, 14, 66-69.	1.7	13
126	Epigenetic profiling of Italian patients identified methylation sites associated with hereditary transthyretin amyloidosis. <i>Clinical Epigenetics</i> , 2020, 12, 176.	4.1	13

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127	Real-life experience with inotersen in hereditary transthyretin amyloidosis with late-onset phenotype: Data from an early-access program in Italy. <i>European Journal of Neurology</i> , 2022, 29, 2148-2155.	3.3	13
128	SOD1 G93D mutation presenting as paucisymptomatic amyotrophic lateral sclerosis. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2009, 10, 479-482.	2.1	12
129	D11Y SOD1 mutation and benign ALS: A consistent genotype-phenotype correlation. <i>Journal of the Neurological Sciences</i> , 2011, 309, 31-33.	0.6	12
130	A novel compound heterozygous <i>ALS2</i> mutation in two Italian siblings with juvenile amyotrophic lateral sclerosis. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2013, 14, 470-472.	1.7	12
131	Sural nerve biopsy in peripheral neuropathies: 30-year experience from a single center. <i>Neurological Sciences</i> , 2020, 41, 341-346.	1.9	12
132	Restless Leg Syndrome in Different Types of Demyelinating Neuropathies: A Single-Center Pilot Study. <i>Journal of Clinical Sleep Medicine</i> , 2013, 09, 945-949.	2.6	12
133	A novel L67P SOD1 mutation in an Italian ALS patient. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2011, 12, 150-152.	2.1	11
134	Letter: faecal microbiota transplantation in combination with fidaxomicin to treat severe complicated recurrent <i>Clostridium difficile</i> infection. <i>Alimentary Pharmacology and Therapeutics</i> , 2015, 42, 1030-1030.	3.7	11
135	Targeting S100A4 with niclosamide attenuates inflammatory and profibrotic pathways in models of amyotrophic lateral sclerosis. <i>Journal of Neuroinflammation</i> , 2021, 18, 132.	7.2	11
136	Neuropathy with Predominant Small Fiber Involvement Associated with Abnormal Anti-MAG Titer. <i>Internal Medicine</i> , 2010, 49, 2627-2629.	0.7	10
137	Pathological Findings in Chronic Inflammatory Demyelinating Polyradiculoneuropathy: A Single-Center Experience. <i>Brain Sciences</i> , 2020, 10, 383.	2.3	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. <i>Neurological Sciences</i> , 2022, 43, 1359-1364.	1.9	10
139	Gastrointestinal Manifestations in Hereditary Transthyretin Amyloidosis: a Single-Centre Experience. <i>Journal of Gastrointestinal and Liver Diseases</i> , 2020, 29, 339-343.	0.9	10
140	Intravenous Immunoglobulin Treatment in Autoimmune Neurological Disorders—Effects on Quality of Life. <i>Human Immunology</i> , 2005, 66, 417-421.	2.4	9
141	Progressive multifocal leukoencephalopathy in a patient with Franklin disease and hypogammaglobulinemia. <i>Journal of the Neurological Sciences</i> , 2009, 284, 203-204.	0.6	9
142	ATTRv in Lazio-Italy: A High-Prevalence Region in a Non-Endemic Country. <i>Genes</i> , 2021, 12, 829.	2.4	9
143	Teaching Neuro <i>Images</i> : The full-blown neuroimaging of Wernicke encephalopathy. <i>Neurology</i> , 2009, 72, e115.	1.1	8
144	Gadolinium enhancement of the lumbar leptomeninges and roots in a case of ALS. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2010, 11, 412-413.	2.1	8

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145	Clinical, electrophysiological and pathological findings of a patient with CMT2 due to the p.Ala738Val mitofusin 2 mutation. <i>Journal of the Neurological Sciences</i> , 2011, 307, 168-170.	0.6	8
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